

**Personal Experiences of Leaders of Family-Led Organizations and their Role as
Advocates: A Thematic Analysis**

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Abstract

There are several different types of family-led organizations that provide support for family members of children and youth with special health care needs, CYSHCN. Family-to-Family (F2F) groups and State Affiliate Organizations (SAOs) are state level organizations that are part of the larger, national Family Voices organization, and focus on providing support related to healthcare systems. Parent-to-Parent groups are also state level organizations that are part of the larger, national Parent-to-Parent organization, and focus on providing one-on-one emotional support. Family-led organizations work at both individual and systems levels to support family members of CYSHCN and to ensure the family voice is included in the care of CYSHCN. All staff of family-led organizations are by definition family members of CYSHCN or CYSHCN self-advocates.

The National Coordinating Center (NCC) and its Regional Genetics Networks (RGNs) work with family-led organizations to help individuals, particularly those in underserved populations, access genetic services. The New York Mid-Atlantic Consortium (NYMAC) is the RGN for the region including Delaware, Maryland, Pennsylvania, New Jersey, New York, Virginia, West Virginia, and the District of Columbia. The family-led organizations within this

region were identified by the National Genetics Education and Family Support Center, an initiative run by Genetic Alliance, which works to support individuals with diverse healthcare needs.

For this study, six leaders of family-led organizations within the NYMAC region were interviewed using a semi-structured interview guide. The interviews were recorded, transcribed, and analyzed using thematic analysis. The three themes and the associated subthemes that were identified include the experience of being a family member of a CYSHCN and the associated difficult emotions; working with providers and expectations and hopes for the CYSHCN, care coordination, and experiences with genetics; and lastly, the experience of being a leader in a family-led organization and issues of access, the progression of the family-led organizations, and ideological differences among family-led organizations. The information and expertise of both the family-led organizations and the leaders of these organizations have the potential to improve the care of family members of CYSHCN and CYSHCN themselves across a variety of systems, which could have a significant impact on public health.

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Preface

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Abbreviations

| | |
|----------|--|
| ACA | Affordable Care Act |
| ACMG | American College of Medical Genetics and Genomics |
| ADA | Americans with Disabilities Act |
| ANOVA | Analysis of Variance |
| CF | Cystic Fibrosis |
| CHIP | Children's Health Insurance Program |
| CMC | Children with Medical Complexity |
| CSHCN | Children with Special Health Care Needs |
| CYSHCN | Children and Youth with Special Health Care Needs |
| D/HH | Deaf or Hard of Hearing |
| DS | Down Syndrome |
| EHDI | Early Hearing Detection and Intervention |
| F2F | Family-to-Family |
| FAPE | Free and Appropriate Public Education |
| FCC | Family-Centered Care |
| GARD | Genetic and Rare Diseases Information Center |
| HCBS | Home and Community Based Services |
| HRSA | Health Resources and Services Administration |
| ID/DD | Intellectual Disability or Developmental Disability |
| IDEA | Individuals with Disabilities Education Act |
| IEP | Individual Education Program |
| KIPP | Kansas Inventory of Parental Perceptions |
| LRE | Least Restrictive Environment |
| MCH | Maternal and Child Health |
| MCHB | Maternal and Child Health Bureau |
| MUA/P | Medically Underserved Areas/Populations |
| NCC | National Coordinating Center |
| NS-CSHCN | National Survey of Children with Special Health Care Needs |
| NYMAC | New York Mid-Atlantic Consortium |
| OBRA | Omnibus Budget Reconciliation Act |
| P2P | Parent-to-Parent |
| PCES | Parent Coping Efficacy Scale |
| PTI | Parent Training and Information Center |
| PTSD | Posttraumatic Stress Disorder |
| RGN | Regional Genetics Network |
| SAO | State Affiliate Organization (of Family Voices) |
| SSDI | Social Security Disability Insurance (Benefits) |
| SSI | Supplemental Security Income |

1.0 Introduction

Family-led organizations provide important tailored support to family members of children and youth with special healthcare needs, or CYSHCN, as well as to the CYSHCN themselves.^{1,2} These family-led organizations help family members of CYSHCN navigate healthcare and education systems, as well as others, and work to ensure access to support.³ All staff of these family-led organizations are by definition one of the following: parents of CYSHCN, siblings of CYSHCN, or CYSHCN self-advocates. There are multiple types of family-led organizations, including Family-to-Family (F2F), Parent-to-Parent (P2P), and State Affiliate Organizations (SAOs) of the national Family Voices organization. Each type has its own specialty; F2Fs focus on healthcare issues and P2Ps focus on matching family members one-on-one with other supporting family members. SAOs work on promoting the mission of Family Voices,³ which is to keep the family voice at the center of the care for CYSHCN in healthcare systems and to advocate to improve policies for children.⁴

There have been several studies analyzing barriers and challenges to achieving quality care,^{5,6} as well as statements detailing the efforts of family-led organizations^{3,7} and other organizations^{8,9} to overcome these barriers, including poor care coordination and the care of children and youth with high levels of special health care needs. There have also been several studies analyzing the positive effects of being connected with family-led organizations,^{10,11} but these have been most often from the perspective of parents of CYSHCN seeking support from the family-led organizations.^{1,2,12-14} There is limited literature on the study of genetic support groups^{15,16} and family-led organizations¹⁷ from the perspective of the leaders within those organizations. These studies are described below. This current study adds both an increase in the

scope of this area of research, as well as the acquisition of new insight from the leader's perspectives.

The medical home model, endorsed by the American Association of Pediatrics, emphasizes continuous, family-centered, coordinated care.⁸ Care coordination is the process by which patients and their families are connected with services in a centrally managed, logical fashion.¹⁸ Beene-Harris, et al. identified barriers to genetic services that included both individual and institutional barriers, the latter including issues with care coordination.⁵ Aspects of care coordination that were discussed by Beene-Harris, et al. included access to information, follow-up care, referral to support groups, assistance transitioning from one stage to another, and quality education.⁵ Family-led organizations not only help family members of CYSHCN and CYSHCN themselves navigate the healthcare system, but they also work at the systems level to advocate for family-centered care, or FCC.⁷

Kuo performed secondary analysis on the 2005-2006 National Survey of CYSHCN and reported on the burdens of care of children with medical complexity (CMC), a more complex subset of CYSHCN.⁶ The findings of Kuo, which covered the degree of success in accessing particular services, reported widespread difficulty in systems navigation and the importance of care coordination for CMC.⁶ A two-part study by Henderson, et al. (2014) and Henderson, et al. (2016) that focused on parents of children who were deaf or hard of hearing (D/HH) analyzed the conceptual framework of the parent-to-parent support connection and identified several integral components unique to this type of support.^{10,11}

The positive effects of the support offered by family-led organizations has been documented largely from the perspective of the parents of CYSHCN. A two-part study by Ainbinder, et al. and Singer, et al. performed both qualitative and quantitative analysis of P2P

support and generated the first empirical data regarding the benefits of this distinctive type of assistance, including improvements in acceptance, coping, and maintaining a positive perspective.^{1,2} Other studies that performed qualitative analysis of interviews with parents included a study done by Kerr, et al. interviewing parents of children with a congenital upper limb deficiency,¹² and a study done by Konrad interviewing parents of seriously ill and dying children,¹³ both of which found that parent-to-parent support was considered helpful. Mathiesen, et al. interviewed and conducted focus groups with parents of children registered with the Utah Birth Defect Network to determine how parental support would affect parents of a child with a structural birth defect. The authors ultimately recommended that parent support groups be associated with birth defect registries throughout all hospitals to provide support for parents of children with structural birth defects.¹⁴

Two studies by Black, et al. and Weiss analyzed a survey of directors of genetic support groups in order to describe the services provided by, and importance of these organizations.^{15,16} Bradham, et al. conducted a study that surveyed state coordinators of early hearing detection and intervention programs in an effort to improve program effectiveness and infant outcomes; the findings supported the use of parent support groups that staffed paid parents of children who were D/HH.¹⁹ A single study that reported on family-led organizations from the perspective of the leaders of these organizations was found. Behl, et al. performed this study in order to understand and improve services for families of children who are D/HH.¹⁷ Room for improvement was found in increasing the percentage of families with children who were D/HH who were accessing family-led organizations, but the benefits of the family-led organizations were also emphasized.¹⁷

This current study aimed to further the scope of this limited area of research by interviewing leaders of family led organizations. These leaders have a wealth of knowledge

applicable to integrating family-led organizations into systems of care and incorporating the medical home model, as well as a unique perspective on the care of CYSHCN. The purpose of these interviews was to explore the leaders' backgrounds, stories, goals, challenges, and evolution as family members of CYSHCN, and as leaders of family-led organizations, where they represent themselves directly. The interview questions asked about the leaders' experiences with and perceptions of access to services, including genetic services. They also addressed how leaders have participated in their organizations and how they have supported individuals through their organizations over time, including specific outcomes. This study sought to explore the barriers experienced and addressed by leaders of family-led organizations, which may also be faced by the general population. The study findings can be used in the future to increase the understanding about, and improve access to, parental support and both healthcare and genetic services.

For this study, interviews with leaders of family-led organizations were conducted and analyzed using thematic analysis. These individuals are leaders of family-led organizations within the New York Mid-Atlantic Consortium (NYMAC) Regional Genetics Network region. The NYMAC region includes Delaware, Maryland, New Jersey, New York, Pennsylvania, Virginia, West Virginia, and the District of Columbia.²⁰ NYMAC is one of seven regional genetics networks (RGNs) that aim to improve access to genetic services across the country, specifically for underserved populations.²¹

1.1 Specific Aims

The aims of this current study are as follows:

Specific Aim 1 Collect the stories, challenges, outcomes, and goals of leaders of family-led organizations by interviewing leaders in SAOs, as well as P2P and F2F organizations.

Specific Aim 2 Analyze the interviews by thematic analysis to

- a) determine how these leaders have participated in these organizations and supported their members.
- b) elicit these leaders' perceptions of genetic services and access to them.

2.0 Literature Review

2.1 Family-Led Organization and Partner Organization Structure

Family-led organizations provide support to families of CYSHCN.^{1,2} CYSHCN are defined as “those who have or are at increased risk for a chronic physical, developmental, behavioral, or emotional condition and who also require health and related services of a type or amount beyond that required by children generally.”^{9(p138)} Family-led organizations work at the individual level to assist with systems navigation and provide one-one-one support, as well as at the systems level to advocate for change, build leadership capabilities, and work with other organizations including the Regional Genetics Networks.³

Under the umbrella of family-led organizations, there are several types that differ in how they work to meet the needs of families of CYSHCN. F2F groups and SAOs are part of the Family Voices network, a national organization that focuses on keeping the family voice at the center of healthcare.³ The national Family Voices organization was first funded in 1993 by the Maternal and Child Health Bureau (MCHB) to support families of CYSHCN and continue to develop family leaders, and the first six F2F groups were piloted in 1999 with additional funding by the Robert Wood Johnson Foundation.²² Its mission statement reads, “Family Voices is a national organization and grassroots network of families and friends of children and youth with special health care needs and disabilities that promotes partnerships with families – including those of cultural, linguistic, and geographic diversity – in order to improve healthcare services and policies for children.”⁴

F2Fs are state level groups that receive funding through the national MCHB of the Health Resources and Services Administration (HRSA), but focus on directly helping families of CYSHCN make use of the healthcare system and identify outside support, such as community, disability, and informational services.^{3,23} SAOs are also groups that function at the state level to advance the mission of Family Voices, and also offer training in working with healthcare professionals and systems to family leaders and professional partners.^{3,23}

Parent-to-parent (P2P) USA is a national network that also has groups that function at the state level.¹¹ P2P groups have existed since 1973, but received their first funding for national efforts in 2003 by the Robert Wood Johnson Foundation; they continue to be funded through other donations.^{17,24} P2P groups connect supporting parents with learning parents, where a supporting parent is defined as a parent experienced in navigating systems and finding support for the care of their CYSHCN and who is ready to partner with other parents of CYSHCN, and a learning parent is defined as a parent new to being a caregiver of a CYSHCN.¹¹ Leaders in all the aforementioned family-led organizations have personal experience either as a parent/guardian or as a sibling of a CYSHCN, or self-advocate CYSCHN. Staff at these family-led organizations can leverage their lived experience to direct incoming families to the organizations that will most benefit them.³ While F2F organizations and SAOs focus directly on healthcare, P2P organizations focus on emotional support for the family members.³ In some states, combinations of family-led organizations are housed at the same location. Table 1 below shows the structure of P2Ps, F2Fs, and SAOs in the region.

Table 1 Family-Led Organizations (P2P, F2F, & SAO only) in Region, adapted.

| State | Subset of Family-Led Organizations in the NYMAC Region | | |
|----------------------|--|--|--|
| | P2P | F2F | SAO |
| Delaware | Delaware Family Voices | | |
| Maryland | Parent to Parent of Maryland at the ARC MD | Parents' Place of Maryland, Inc. | |
| New Jersey | Statewide Parent Advocacy Network (SPAN) | | |
| New York | Parent to Parent of NYS | | |
| Pennsylvania | Parent to Parent of PA | Parent Education and Advocacy Leadership Center (PEAL) | |
| Virginia | Center for Family Involvement | Family to Family Network of Virginia - Center for Family Involvement | |
| West Virginia | | West Virginia University's Center for Excellence in Disabilities | West Virginia Parent Training and Information |
| District of Columbia | | Advocates for Justice and Education | Family Voices of the District of Columbia Inc. |

*Note: Adapted from Referral to Support and Partnering with Family-Led Organizations.³

A diagram illustrating the relationships of these family-led organizations to each other, as well as to their partner organizations, is shown in Figure 1. These partner organizations, which are discussed below, include national and regional organizations that focus on genetic and healthcare services.

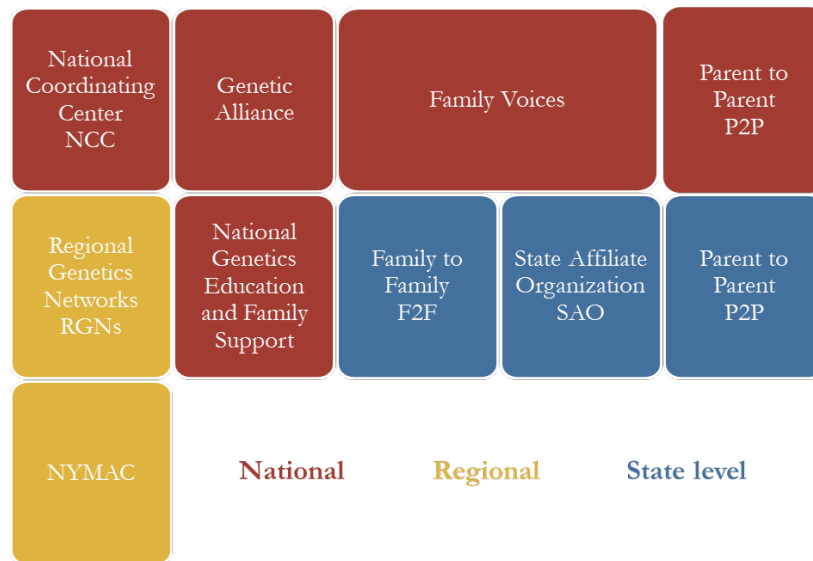


Figure 1 Diagram of Subset of Family-Led and Partner Organizations at the National, Regional, and State Level.

One group working to bring together leaders of family-led organizations, healthcare professionals, and service delivery specialists is the Regional Genetics Networks and their National Coordinating Center (NCC). The United States was divided geographically in 2017 into seven Regional Genetics Networks (RGNs) and each RGN was awarded a grant by the MCHB of HRSA.^{21,25} The NCC and the National Genetics Education and Family Support Center were also each awarded a grant. The organizations receiving these grants were charged with linking advances in genetics with improvements in public health.²⁵ These three organizations have a joint mission that states, “The mission of the seven Regional Genetics Networks, the National Coordinating Center for the Regional Genetics Networks, and the National Genetics Education and Family Support Center is to improve access to quality genetic services for medically underserved populations.”²¹

The National Genetics Education and Family Support Center is a three-year initiative that works alongside the RGNs to connect individuals and families to direct support. This Family Support Center helps the RGNs to partner with family-led organizations and reach underserved

families, provide information on education and healthcare for genetic conditions, connect families with emotional support and other services, and provide training opportunities at the individual and systems level.³

Family-led organizations not only work to directly help family members of CYSHCN and CYSHCN themselves, but also work to advocate for family-centered care (FCC), which emphasizes partnership between the family and the health professional.⁷ Genetic Alliance is the national organization that is leading the Family Support Center initiative, as well as working on advocacy-led research, family planning, and stakeholder engagement programs.²⁶ While Genetic Alliance was originally focused on supporting individuals with genetic conditions, the organization has expanded to supporting individuals and families with diverse healthcare needs and endeavors to keep them active participants in healthcare.²⁷ Their mission statement reads, “Genetic Alliance engages individuals, families, and communities to transform health.”²⁸

NYMAC, or the New-York-Mid-Atlantic Consortium Regional Genetics Network, is the Regional Genetics Network for Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia. NYMAC’s current goals include increasing “the number of individuals or families within the geographic area served by the RGN, the number of medically underserved patients served by each RGN, the number of primary care providers using RGN resources, the percentage of clinical sites that use telehealth/telemedicine to provide genetic services, and the number of medically underserved patients receiving genetic services through telemedicine visits.”²⁹ Like F2Fs, NYMAC is funded by the Maternal and Child Health Bureau through HRSA.

2.2 The Context of Family-Led Organizations

Over time, many individuals, groups, and organizations have worked and collaborated in order to achieve the current state of family-led organizations and care for CYSHCN; there have been several milestones accomplished through, and related to, this work. During the 1980s and 1990s, a series of Surgeon General's conferences were held in conjunction with the MCHB and were integral in bringing families together to discuss strategies in order to improve services.²² The MCHB then began funding family-led organizations' projects, which connected these organizations to overarching Maternal and Child Health (MCH) initiatives.²² After the Omnibus Budget Reconciliation Act (OBRA) of 1989 emphasized using what would come to be known as the medical home model in the care of CYSHCN, the partnership between the MCHB and family-led organizations was solidified.²² Since then, the MCHB has funded Family Voices and many F2Fs.²² Additionally, state Title V MCH programs have worked consistently with family-led organizations.²² Where family-led organizations were originally involved only in state CYSHCN programs, they have since been integrated into broader state MCH programs.²² Table 2 lists several of the events related to both the input of, and the development of family-led organizations.

Table 2 Timeline of Key Events Supporting CYSHCN, adapted.

| Year | Event |
|---|---|
| 1935 (Amended) | Social Security Act Included Title V Maternal and Child Health Services Block Grant Program Awards funds to states for healthcare services for mothers, infants, and children, including CYSHCN ³⁰⁻³² |
| 1965 | Medicaid enacted as Title XIX of the Social Security Act ³³ Gives states option of receiving funding for health care services for low-income children, the blind, and individuals with disabilities |
| 1975 | The Education for All Handicapped Children Act – PL 94-142 ³³ |
| 1981 (Authorized 1984) ³⁴ | The Omnibus Budget Reconciliation Act of 1981 – PL 97-34 ³³ Home and Community Based Services (HCBS) Medicaid Waiver Program Supports families to care for their CYSHCN at home by allowing states to fund home and community services |
| 1982-1992 | Surgeon General’s series of six conferences ²² MCHB and Surgeon General hold conferences that incorporate families into partnership for improving services |
| 1984 | Katie Beckett granted individual exception (waiver) by Reagan administration ³⁵ Allowed Katie to be cared for at home instead of an institution |
| 1989 | OBRA of 1989 – PL 101-239 ³³ Amended Title V Maternal and Child Health Services of the Social Security Act to emphasize “family centered, community based, coordinated care” ^{22(p254)} for CYSHCN |
| 1990 | Americans with Disabilities Act (ADA) ³⁶ Protects individuals with disabilities from discrimination based on their disability |
| 1990 | Individuals with Disabilities Education Act (IDEA) – PL 94-142 ³⁷ Name changed from The Education for All Handicapped Children Act Focuses on individual, designed to ensure equal education opportunities to children with disabilities |
| 1993 | MCHB funds Family Voices to support families of CYSHCN and develop network of family leaders ²² |
| 1998 | Formal definition of CYSHCN published in <i>Pediatrics</i> ^{9,33} |
| 1999 | OLMSTEAD v L.C. 98-536 527 US 581 ³³ Supreme Court upholds ruling that individuals with disabilities have the right to live in their communities based on provisions of the ADA, ³⁶ leads to adoption of HCBS waivers by states |
| 1999 | F2Fs piloted in 6 states by Family Voices, supported by MCHB and funded by Robert Wood Johnson Foundation ²² |
| 2002 | MCHB awards grants to 6 states to develop state-wide F2Fs ²² |
| 2005 | MCHB funds F2Fs in every state ²² |
| 2010 | The Patient Protection and Affordable Care Act (ACA) – PL 111-148 ³³ Ends exclusions for preexisting conditions as well as annual and lifetime caps for coverage |

*Note: Adapted from Allshouse, et al. and Reynolds, et al.^{22,33}

The Individuals with Disabilities Education Act, or IDEA, of 1990 updated the Education for All Handicapped Children Act to focus more on the individual, instead of their diagnosis, while maintaining the purpose of ensuring an appropriate education for children with disabilities.³⁷ There are six pillars of IDEA: a child has a basic right to a free and appropriate public education (FAPE); a child will have an evaluation for disability if indicated by an educational professional; a child must be educated in the least restrictive environment (LRE), or as close to the environment of a typical child as possible; the child, parents, and teacher must work together regarding the child's education; each child will have an Individual Education Program (IEP) developed for them to plan for their success; and the parents of the child are guaranteed a list of rights regarding their child's education, for example, access to their child's records.³⁸ An IEP is a binding document put together by a team of individuals including the teacher, a counselor, an administrator, a special education teacher, the child, and the parents of the child, and may include others. IEPs delineate plans to reach the goals of the child, and state specific actions and steps designed to achieve that plan.³⁸

Parent Training and Information Centers (PTIs) are another type of family-led organization that focus on assisting families of CYSHCN with issues related to education, including early intervention.³ PTIs are commonly contacted by families of CYSHCN regarding issues related to IEPs. In many states, one organization houses the F2F and the PTI, and staff may be cross-trained to address both healthcare and education questions. As such, while this study did not include speaking specifically with leaders of the PTIs, both IDEA and IEPs are addressed on a regular basis by staff of family-led organizations including F2Fs.

One current reform in the care of CYSHCN is, in part, including not only children who are already identified as having a special health care needs, but also at-risk children. A work group

convened to establish the definition of CYSHCN by the MCHB's Division of Services for Children with Special Health Care Needs included at-risk children in the definition of CYSHCN in order to aid in planning and development efforts for preventative strategies.⁹ At-risk children may include those with certain biological or environmental risks, such as very low birth weight or extreme poverty.⁹ The work group cited two reasons for including at-risk children in the definition of CYSHCN.⁹ The first being that there is scientific reasoning behind it, as there are multiple determinants of health, including biological, social, and environmental, that result in a continuum of the quality of children's health.⁹ Secondly, while prevention may reduce the future needs for specialized healthcare, there has been a lack of prevention efforts targeted to at-risk populations of children.⁹ The work group chose to view this history as an impetus to embrace the task, instead of avoid it.⁹

A major focus of the current reform movement in the care of CYSHCN is providing family-centered care,⁷ as outlined in the OBRA of 1989 federal legislation.²² Family-centered care is one component of the medical home model endorsed by the American Academy of Pediatrics.³³ The medical home model was originally proposed for the care of CYSHCN, but its use has expanded to the care of all children; it outlines care that is also coordinated, accessible, consistent, and culturally appropriate.³³ The American Academy of Pediatrics Policy Statement updating its 1992 operational definition of medical home adds that it should be delivered by a physician who knows the family professionally, works with them to direct the care of their child, acts as a primary care physician, and coordinates care.⁸ Care coordination is the coordinated effort of connecting patients and their families with services to ensure quality healthcare.¹⁸

An integral component to the success of family-centered care is the acknowledgement that the caregivers of CYSHCN are the experts on the person for whom they are providing care.^{33,39}

Caregivers, including mothers, fathers, grandparents, siblings, and guardians, commit to intensive, full-time, long-term care. These caregivers fulfill many roles, including performing clinical tasks, managing home care providers, researching necessary information, keeping track of reports and budgets, finding needed services, and coordinating their child's schedule, in addition to managing their other responsibilities and goals.³⁵ Caregivers see the child as a whole person and often conduct care coordination directly for them. It is in this way, through their own initiative, that most leaders of family-led organizations begin their journey.²²

Upon learning that their child has a special health care need, family members of CYSHCN have different needs, which change over time. Individuals may seek out information about understanding their child's medical condition or finding available services, and/or they may seek out emotional support from others who have been in similar situations.^{3,22} In this way, family members of CYSHCN can be connected with family-led organizations in their state. These organizations can help families navigate the systems of care, including health care, education, and insurance issues.³ Additionally, staff at these organizations represent multiple cultures and often speak different languages, strengthening the connection of new parents.³

Family members learn advocacy, leadership, and communication skills on behalf of their child, and then bring this expertise to the family-led organization, where over time they may begin to support other families in the community.²² Family members of CYSHCN may become involved by sitting on boards, representing the family perspective in procedures such as focus groups or surveys, or connecting with other volunteer opportunities.²² As family members continue to advocate for their own child, they often recognize the impact they may have by advocating for all CYSHCN at the systems level. These family members may become involved with services, policy, and program efforts, as they are supported by their peers in the family-led organizations.²² Family

members may also become involved at the systems level by providing testimony at hearings, and writing editorial letters and letters to government officials.²² Many family members of CYSHCN already advocating for their own, and other's children are invited to work for, as well as to become leaders in family-led organizations. Family-led organizations endeavor to have family members of CYSHCN involved at the individual, community, and systems levels.

2.3 Barriers to Access to Quality Healthcare for CYSHCN

2.3.1 Geographical, Cultural, and Financial Barriers to Healthcare Services

Family-led organizations work to help families achieve access to services and programs, as well as overcome barriers in areas such as insurance and education. Family members of CYSHCN may find themselves struggling with finding an appropriate specialist or other healthcare providers, especially if they are in a remote area. Specialists needed by CYSHCN are concentrated at specific institutions that are not evenly geographically distributed.³³ These specialists are scarce, and the family's location may considerably limit their access to care.³³ Additionally, literacy, language, and cultural barriers can also impede access to needed services or programs.^{35,39} Family members with reduced literacy or English proficiency may face obstacles in healthcare decision-making, for example, in regards to determining the overall cost of their child's care under a particular insurance policy.³⁵ Difficulties with health insurance and financing medical and therapy needs are widespread barriers for family members of CYSHCN.^{33,40} Even families with premium private insurance plans may still incur substantial out-of-pocket costs.³³

2.3.2 Limited Utility of Governmental Assistance Programs

The linkage of Medicaid with individual states can limit where a CYSHCN seeks treatment and where family members seek employment.³³ As Allshouse, et al states, “State-specific eligibility limits and Medicaid waiver waitlists represent significant barriers to coverage and, by extension, to care.”^{33(pS198)} The HCBS Medicaid waiver program that was passed in 1981 and authorized to apply to disabled children in 1984,³⁴ allows states to fund the care of CYSHCN in their home and community settings as opposed to an institutional setting.³³ The term “waiver” derives from the waiver of certain Medicaid rules (the requirement of care provided in an institutional setting) for individuals to be eligible for this benefit.⁴¹

The initial uptake of the Medicaid waiver program by states was slow, but subsequent legislation made it easier for states to adopt the program.³⁶ Additionally, the ruling in the court case of *OLMSTEAD v L.C.* in 1999 greatly increased the adoption of the Medicaid waiver program by states, as the Supreme Court ruled that unnecessarily institutionalizing people with disabilities violated their civil rights as outlined in the ADA of 1990.³⁶ This ruling led to the development of guidelines and grants to assist states in complying with this Supreme Court case.³⁶

In spite of these events, adoption of Medicaid waivers by states remains optional, states determine the eligibility criteria, and each state determines the maximum number of enrollees it will allow.⁴⁰ The cap on the number of individuals receiving a Medicaid waiver results in a waitlist for each state.⁴⁰ These waitlists are not exclusive to CYSHCN and individuals can be on the waitlist for a substantial amount of time. In 2017, over 707,000 people across 40 states were on a waitlist for a HCBS Medicaid waiver.⁴⁰ The average wait time in 2017 for these waivers was 30 months, but the waivers (and their wait times) differ by target population.⁴⁰ The average wait times for these waivers ranged from four months for those with HIV/AIDS to 66 months for those

with intellectual disability or developmental disability (ID/DD).⁴⁰ One family, mentioned by Allshouse, et al. to illustrate the shared family experience, had been (and continued to be) on the Medicaid waiver waitlist for 12 years.³³

Other federal assistance programs include Social Security Disability Insurance/Benefits (SSDI), which provides funds to disabled individuals under 65 years of age who have worked long enough, and Supplemental Security Income (SSI), which provides funds to disabled children and adults with minimal income. To be eligible for SSDI, an individual (or one of their parents) must have worked for a certain number of years depending on the age at which they become disabled and the individual must also be under 65 years of age.⁴² Additionally, the individual must have a medical condition predicted to last for at least 12 months that prevents the individual from working or that results in death. SSI eligibility does not require work history but does require minimal income.^{43,44} Additionally, the individual must have a medical condition that meets the same criteria as SSDI eligibility (or be over the age of 65). While these programs exist, CYSHCN and their family members may find that their applications are not accepted. In outlining the details of SSDI and SSI for those who may need to apply due to the difficulties associated with a mental health condition, the National Alliance on Mental Illness concludes the webpage on accessing support with information about appealing a denied claim, stating, “the majority of claims are denied, and most people file an appeal.”⁴⁵

2.3.3 Outcome Measures: Partners in Shared Decision-Making and Receipt of Care in a Well-Functioning System

While there are substantial geographical, cultural, and financial barriers to obtaining quality healthcare for CYSHCN, there are also other widespread barriers faced by families of

CYSHCN.³³ According to data from the 2009/2010 National Survey of Children with Special Health Care Needs (NS-CSHCN), which surveys families of non-institutionalized CSHCN between the ages of 0 and 17, 15.1% of children in the nation have special health care needs, which is an estimated 11,203,616 children.⁴⁶ This survey records information on the impact on the family of having a child with special health care needs, and reports that 22.1% of families with CSHCN pay \$1,000 or more out of pocket on the child's medical expenses per year.⁴⁶ Additionally, 13.1% of families with CSHCN report spending 11 or more hours per week providing or coordinating the child's healthcare.⁴⁶

This survey also reports on core outcome measures, which include the percentage of families with CSHCN that are partners in shared decision-making for the child's optimal health.⁴⁷ The data in this outcome measure are also broken down by race and ethnicity.⁴⁷ The following are the percentages of families where this outcome was not achieved: Hispanic 36.5%, Black non-Hispanic 35.3%, Other non-Hispanic 33.2%, and White non-Hispanic 25.8%.⁴⁷ This survey shows that there are significant impacts on families of CSHCN and that those impacts are not experienced equally across all subpopulations.

The NS-CSHCN also reports aggregate data on whether national performance and outcome measures were achieved.⁴⁸ One such Title V Maternal and Child Health Block Grant National Outcome Measure reported is that CSHCN received care in a well-functioning system, as measured by the achievement of all six core outcomes.⁴⁸ This national outcome measure is made up of five core outcomes for children 0-1 and six core outcomes for children 12-17.⁴⁸ The first five core outcomes include family partnership, medical home, early screening, adequate insurance, and easy access to services.⁴⁸ The additional core outcome (for a total of six) for children 12-17 is preparation for adult transition.⁴⁸ The 2009-2010 NS-CSHCN reported that only 17.6% of

CSHCN achieved all of their age-relevant core outcomes.⁴⁸ The amount of CSHCN who did not achieve one age-relevant core outcome was 24.5%, and 57.9% of CHSHCN did not achieve two or more age-relevant core outcomes.⁴⁸

Additionally, each core outcome incorporates the measurement of multiple variables.⁴⁹ The fifth core outcome, easy access to services, is made up of six variables that are each difficulties with accessing care, including not eligible for services, services not available in your area, waiting lists or other problems getting appointments, issues related to cost, trouble getting the information needed, any other difficulties not mentioned.⁴⁹ To not achieve the core outcome of easy access to services, at least one of these six difficulties had to be present and the family must have reported that they were usually or always frustrated in their efforts to get services.⁴⁹ The amount of CSHCN reported to not have achieved this specific core outcome of easy access to services was 34.9%.⁴⁹

2.3.4 System Fragmentation as a Barrier to Healthcare Services

The fragmentation of the healthcare system further complicates the treatment of CYSHCN,^{9,36} and generally places the burden of vital care coordination on parents of CYSHCN.³³ Fragmentation is present both within systems, such as when healthcare provider offices do not communicate with one another, and between systems, such as when healthcare issues are not communicated to educational institutions. The medical home model, which incorporates care coordination and family-centered care, is one way to address this problem but is not currently widely adopted.³³ Fragmentation affects healthcare professionals and family members alike in that they both have to work to orchestrate the care of the CYSHCN. But, the disconnected relationship of systems guarantees that families best understand the complexities of fragmentation when they move within and between these systems, as the family members are the constant in the

life of the CYSHCN.³³ In addition to time and money, there are other resources that families of CYSHCN must carefully manage. Additional quality markers for families of CYSHCN include minimizing school and work days missed as well as hospital admissions, and ensuring wage and job stability.³⁵

2.3.5 Individual and Institutional Barriers to Accessing Genetic Services

One study by Beene-Harris, et al. investigating barriers to accessing genetic services analyzed data from five focus groups.⁵ The five focus groups were organized by target subpopulations: a sickle cell anemia parent support group, a Native American student group, parents of children with birth defects or other special health care needs, adults with genetic conditions, and genetic counselors from the state of Michigan, and totaled 48 individuals.⁵ After content analysis of the interview transcripts was completed, a series of barriers were identified, either on the individual or institutional level.⁵

Barriers identified on the individual level included lack of awareness of risk, lack of knowledge and awareness of genetic services or resources, and lack of trust and fear of discrimination.⁵ Regarding the last barrier, one participant diagnosed with neurofibromatosis said, “How can I expect the world not to discriminate against my son when people don’t even tell family members that there’s something wrong? And if doctors tell you not to tell anyone. They really scare them. The early statements of doctors really stay with you.”^{5(p13)}

Barriers identified on the institutional level included provider lack of knowledge and awareness of genetic services, lack of workforce, coordination of care/referral, cost and insurance, and distance from services.⁵ Aspects of care coordination that were discussed included access to information, follow-up care, referral to support groups, assistance transitioning from one stage to

another, and quality education.⁵ Support groups (or chat rooms) were described as helpful, and lessened feelings of isolation, for many participants in the adult focus group.⁵ One participant diagnosed with muscular dystrophy said, “I decided to join an internet chat. I thought it would just be people talking trash. But it turned out quite differently. I didn’t think there would be that kind of intimate connection, but it really has been a great help.”^{5(p15)} These statements highlight the considerable need for human connection in those with special health care needs. This connection can take the form of improved communication, immediately available care coordination, and awareness of support groups, among others.

The focus group study was part of a larger needs assessment initiative designed to identify and prioritize genetic health services in the state of Michigan and was utilized to create a framework for the development of the state’s genetic action plan.⁵⁰ This framework listed six goals for public health genetics in the state developed by twelve expert working groups.⁵⁰ One of these goals was to improve access to genetic information, prevention strategies, and services.⁵⁰ The report for this coordinated needs assessment compiled data from many studies, including a state-wide survey of both healthcare providers (n=140) and genetic service providers (n=54).⁵⁰ This survey found barriers to referral or utilization of genetic clinic services included issues with scheduling, cost, distance to clinic, transportation, and a lack of culturally competent education materials.⁵⁰ Other barriers included confidentiality concerns and a fear of knowing genetic test results, as well as lack of public knowledge and patient refusal.⁵⁰

2.3.6 Lack of Awareness of Availability of Genetic Services

A two-part study by Collins, et al. (2001) and Collins, et al. (2003) conducted in Australia first analyzed semi-structured interviews of parents of children with either cystic fibrosis (CF) or

Down syndrome (DS), and secondly performed analysis of surveys of parents of children with DS.^{51,52} In Australia, while there is a program in place for genetic counseling after a CF diagnosis, there is not a program in place for genetic counseling after a DS diagnosis.⁵¹ Four parents of children with CF and ten parents of children with DS were interviewed; six parents of children with DS had received genetic counseling and four had not.⁵¹ Of those who did not have genetic counseling, two parents did not know it was an option, and the other two, while they had vaguely heard of it, did not think they were eligible as they were not yet having another child or they were a single parent.⁵¹ The authors went on to state that there was a need for primary care providers to accurately depict the process of genetic counseling, as well as the reasons for which it can be useful.⁵¹ Of the 74 mothers of children with DS who were surveyed, only 18 had received genetic counseling.⁵² Of the mothers who had not received counseling, most reported that they were not offered it or had never heard of it (71%), and also of the mothers who had not received genetic counseling, but had heard of it, many said they had little to no idea of what genetic counseling was (50%) and they were not sure how genetic counseling could help (73%).⁵² The conclusions in parts one and two of this study align well with each other.⁵² The findings from the qualitative analysis were that the parents did not know genetic counseling was an option, or did not believe they were eligible for it, and these were the main reasons for not having genetic counseling; the finding from the quantitative analysis was that the parents' lack of awareness of the genetic counseling service was a major factor in not having genetic counseling.⁵²

2.3.7 Lack of Awareness of Patients' Need for Referral to Genetic Services

In the Netherlands general practitioners are the “gatekeepers” to clinical genetics; one study by Aalfs, et al. investigated why women would be referred to genetic counseling services

during pregnancy instead of before pregnancy.⁵³ Questionnaires were distributed to general practitioners whose patients had come to the genetic clinic during pregnancy to understand the reasoning of the timing of the referral, the discussion of the referral with the patient, and the knowledge and attitudes towards genetic counseling of the provider.⁵³ Of the referrals, 29% were not completed by the general practitioner, but rather by patients and other medical professionals, and 40% of the referrals were initiated by patient.⁵³ The main reason cited by general practitioners for not referring a patient to prenatal genetic counseling before a pregnancy occurred was that they did not know they should, as they did not know of the potential risk factors present in the patient and/or her family (71%).⁵³ The authors determined (via other survey questions) that a lack of knowledge or acceptance of clinical genetics and genetic counseling was not the reason for the timing of the referral, but rather the lack of the general practitioners' awareness of the patients' needs for referral.⁵³ The authors went on to suggest that general practitioners or other healthcare professionals in the same clinic obtain and routinely update family histories of patients, so as to better know the risk factors present in the patient and/or their family members, as well as to successfully complete referrals for preconception genetic counseling.⁵³

2.3.8 Reviews of Barriers to Accessing Genetic Services

These studies were four of nine reviewed by Delikurt, et al. in 2015 to perform a meta-analysis on the barriers to referral to genetic services (the other five articles were studies regarding cancer-related services).⁵⁴ Thematic analysis of these studies revealed three barriers on the individual level, including lack of awareness of personal risk, awareness of medical history of family members, and knowledge of genetic services.⁵⁴ Six barriers on the (non-genetic) healthcare provider level that are all associated with one another were also identified.⁵⁴ These included lack

of awareness of patient risk factors, not obtaining accurate family history, lack of knowledge of genetics and genetic conditions, and lack of awareness of genetic services, as well as poor care coordination for referrals.⁵⁴ The last barrier identified was the lack of a sufficient genetics workforce.⁵⁴ These barriers span logistical issues, patient education and awareness problems, and trouble with non-genetic healthcare professionals' awareness of genetic services and referral to these services. Partnering with family-led organizations may lessen the magnitude of these challenges.

A previous review by Suther, et al. in 2003 analyzed 18 research studies that identified barriers to the process of primary care providers delivering genetic services, including completing screening tasks, taking family histories, and providing referrals.⁵⁵ Four barriers were identified in three or more articles and included lack of genetic knowledge, lack of detailed or updated family history, lack of referral guidelines or tools to facilitate their use, and lack of confidence for delivering genetic services, assessing, and managing risk.⁵⁵ These barriers are very similar to the ones identified in the 2015 review analyzing non-genetic healthcare provider level barriers previously discussed.^{54,55} This review also evaluated the quality of the included studies, specifically mentioning that only one⁵⁶ of the 18 asked the general providers what their perceived barriers were.⁵⁵ Pediatricians, obstetricians, family medicine doctors, and internal medicine doctors were interviewed for a total of 55 participants.⁵⁶ The perceived barriers to the use of genetic services by these general practitioners included how the test outcome would alter patient management (60%), the sensitivity and specificity of the test (43%), and how much the test costs (42%).⁵⁶

2.3.9 Healthcare Inequities Affecting Underserved Populations Including Racial and Ethnic Minorities

In addition to the barriers to genetic services already listed, healthcare inequities affecting underserved subpopulations⁵⁷ and racial and ethnic minorities⁵⁸ have the potential to increase the disparities in access to genetic services. One study by Suther, et al. (2009) investigated specific barriers to genetic testing by analyzing differences between three racial and ethnic groups: White, Black, and Latino.⁵⁸ The four barriers investigated were knowledge about genetic testing, type of health insurance coverage, concerns about the potential misuse of genetic testing, and lack of trust in a medical doctor to keep their medical information private.⁵⁸ The source of the national self-reported data for 1724 individuals was the 2000 University of Maryland College Park Survey Research Center.⁵⁸ The study found that Blacks and Latinos had less knowledge of genetic testing than non-Hispanic Whites, were less likely to be covered by private health insurance, were more likely to be concerned about misuse of genetic testing, and were more likely to have a lack of trust in a medical doctor.⁵⁸ Suggestions to minimize these disparities included targeted education about genetic testing and its benefits to minority communities and the addressing of cultural issues by healthcare professionals.⁵⁸

An editorial by Goldenberg, et al.,⁵⁷ which preceded two community-engaged studies by the same group,^{59,60} discussed barriers to the use of genetic services by underserved populations. In their editorial, Goldenberg, et al. mention barriers including high cost, lack of insurance plan coverage, lack of the provision of educational services, shortage of the genetic workforce, concerns regarding confidentiality, mistrust of healthcare system and providers, and fears of discrimination.⁵⁷ There are also fundamental barriers to the use of genetic services by underserved populations, including concerns about how the genetic data will be used and integrated with

environmental exposure data, as well as the ability of the underserved communities to leverage economic and structural resources to enact change to reduce the impact of risk factors.⁵⁷

Additionally, it is conceivable that healthcare providers and researchers will make the assumption that underserved populations are not interested in genetic services, which becomes a self-fulfilling prophecy in which underserved communities do not have access to and thus cannot benefit from genetic services.⁵⁷ This then leads to a widening of the gap regarding access to available services between subpopulations.⁵⁷ Suggestions made to prevent this include research within underserved communities regarding barriers to genetic services, and open communication between healthcare providers, researchers, and members of the underserved communities.⁵⁷

The first community-engaged study by this group sought to research perceptions of genetic services and health disparities in underserved individuals in Cleveland, Ohio.⁵⁹ Both a Community Advisory Board and a Network of Community Partners were consulted.⁵⁹ Focus groups were organized by race (African American, Hispanic, and White) and age (below or above age 35).⁵⁹ Flyers and postcards, as well as word-of-mouth recruitment methods were used to enroll 106 participants who completed a total of 13 focus group sessions.⁵⁹ Qualitative analysis of the transcripts resulted in four themes consistent throughout the focus groups: family history and genetic predisposition; genetics, race, and health disparities; addressing multiple determinants of health (including genetics); and social/environmental triggers of genetic traits.⁵⁹ Participants discussed genetic influence accurately but used the language of family history, indicating that this was a good strategy for future communication of genetic predisposition and risk reduction.⁵⁹ This also supports the continuing theme of the importance of taking a family history by healthcare providers.⁵⁹ The participants discussed the different factors affecting the health of individuals in their community, specifically environmental and behavioral and how the factors of environmental,

behavioral, and social influences may trigger a genetic predisposition.⁵⁹ Based on these findings, the integration of gene-environment interactions and the multiple determinants of health in future genetic research was supported, especially in an effort to increase its significance in health disparities research.⁵⁹ Lastly, the importance of including perspectives from individuals in underserved communities in research on understanding and reducing health inequities was emphasized.⁵⁹

The second community-engaged study by this group sought to gain additional insight into the perceptions of genetics and health disparities in underserved populations.⁶⁰ This was done in order to include these individuals in the deliberation on whether genetics research would be relevant to the lives of the underserved, or if it would merely diminish the emphasis on social and environmental determinants of health.⁶⁰ A photo-documentation and reflective writing method was used to both engage the community and share the findings of the study with them.⁶⁰ Two groups of individuals, Latino youths and Africa-American seniors in the Cleveland area, were given training on genetics, health disparities, and photography, as well as the findings from the previously discussed study.⁶⁰ They participated in group discussions before going out into the community to document the aforementioned themes.⁶⁰ Lastly, participants spent one week sharing their photographs with each other and writing about their thoughts on them.⁶⁰ Art openings were held in the communities and attended by participants and community members.⁶⁰ Themes of the work were similar across both groups and included chronic disease, medication, alcohol and drugs, challenges in poor neighborhoods, and the impact of family, socialization, and health beliefs on genetic and socioeconomic factors.⁶⁰ These two community-engaged studies^{59,60} realized the two suggestions presented in the editorial:⁵⁷ to perform research within underserved communities

regarding barriers to genetic services, and to participate in open communication between professionals and members of the underserved communities.

2.3.10 Use of Parent Mentors in Improving Health Outcomes in Minority Children

In the United States the following percentages of children, broken down by race and ethnicity, are uninsured: 5% of White children, 12% of Latino children, 8% of African American children, and 8% of Asian/Pacific Islander children.⁶¹ Many children who are eligible for Medicaid/Children's Health Insurance Program (CHIP) are not enrolled; of those with family incomes below 200% of the federal poverty level, 84% of children are eligible but not enrolled in health insurance.⁶¹

Methods to enroll uninsured minority children in health insurance in 123 families in the Dallas area were compared in a study by Flores, et al. (2016) that examined the effects of pairing the primary caregiver with a parent mentor.⁶¹ The study found that parent mentors were more effective than traditional outreach (as used in the control group of 114 families) and this effectiveness extended to every variable measured, including insuring children, achieving faster coverage, high parental satisfaction, and coverage renewal.⁶¹ Having parent mentors also decreased the likelihood that the children would have no primary care provider, problems getting specialty care, unmet preventative or dental care needs, dissatisfaction with doctors, and need for additional income for medical expenses.⁶¹ Two years after stopping the intervention, 100% of children with parent mentors were still insured.⁶¹ Peer mentors were also found to be more cost-effective than traditional passive methods of Medicaid/CHIP outreach, and it was proposed that peer mentors for adults may have similar benefits in reducing insurance disparities.⁶¹

A previous study by the same group investigated the use of parent mentors in improving asthma outcomes in minority children.⁶² Children with parent mentors (n = 112) had reduced wheezing, asthma exacerbations, and emergency department visits when compared to children in the control group (n = 108) receiving traditional asthma care.⁶²

2.3.11 Subpopulation of Children with Medical Complexity

In addition to underserved and minority subpopulations, barriers to healthcare access of other subpopulations have also been investigated. CYSHCN make up approximately 15.1% of those 18 years and younger in the United States,³⁵ or 11.2 million children. Children with medical complexity (CMC) make up approximately 3.2% of the population of CYSHCN.³³ CMC are a subpopulation of CYSHCN defined as those who are “clinically recognized by at least one chronic condition resulting in high family-identified service need, medical equipment addressing functional difficulties, multiple subspecialist involvement, and elevated health service use”^{6(p1020)} and account for more than one third of the spending on pediatric healthcare³³. CMC are a vulnerable population of more medically complex children that are especially sensitive to the barriers to access to quality healthcare previously described. As such, the demands placed on families of CMC are exceptionally high.

One study by Kuo conducting secondary analysis of the 2005-2006 National Survey of Children with Special Health Care Needs found the median number of physician visits in one year to be 11-15, and the median number of school days missed in the last year to be 10.⁶ Families of CMC were also found to spend a median of two hours providing care coordination per week, and a median of 11-20 hours providing direct home care per week.⁶ The study also found that families of CMC report less likelihood of receiving prescription medications when compared to families of

less complex CSHCN.⁶ The study stated that families of CMC had greater need for care coordination, as they were more disposed to having several unmet service needs, including difficulties accessing nonmedical services.⁶ Table 2 details additional study findings below.

Table 3 Findings of Study Analyzing the 2005-2006 NS-CSHCN, adapted.

| Variable Reported | Percent of Families |
|--|----------------------------|
| Paying more than \$1,000 in out-of-pocket healthcare costs in the prior year | 46.3% |
| Have had healthcare-related financial problems | 56.8% |
| A family member stopped working because of the child's health | 54.1% |
| A family member cut back on working hours to care for the child | 45.6% |
| The family needed additional income for medical expenses | 48.7% |
| Variable healthcare service needs of the child (i.e. needs change all the time or sometimes) | 65.0% |
| At least one unmet medical service need in the last 12 months | 48.8% |
| Five or more unmet medical service needs in the last 12 months | 5.4% |
| Difficulties in accessing nonmedical services | 33.1% |
| Being very satisfied with medical services | 39.4% |

*Note: Adapted from Kuo.⁶

CMC represent a vulnerable subpopulation of CYSHCN, with high levels of need for services, equipment, specialists, and supports. Thus, CMC act as indicators, as their needs make them sensitive to issues in the healthcare system. CMC experience the same barriers to well-functioning care as CYSHCN and children in the general population, including system fragmentation, lack of care coordination, high costs and poor health insurance coverage, issues with obtaining quality home care, and lack of attention to self-care training and the social determinants of health.³³ Allshouse, et al. states, “These deficits point to shortcomings in our health system, but often hit the CMC population first and hardest, making them the equivalent of canaries in the coal mine of America’s evolving healthcare landscape.”^{33(pS196)} While the passage of the ACA has helped families of CMC, they still experience a significant strain on their

resources.³³ Adoption of the medical home model incorporating care coordination, improvements in communication channels, and increased flexibility in available supports is one way to address these issues. This family-centered approach to healthcare would improve access to care for not only CMC, but also other children. As Allshouse, et al. states, “Addressing current deficiencies impacting CMC can result in a similar impact on the pediatric population as a whole.”^{33(pS196)} While the medical home model was originally proposed for the care of CYSHCN, its use has expanded to the care of all children; other advances may also follow this same trajectory.

2.3.12 Involvement of Family-Led Organizations

When reflecting on the origins of FCC, Goldfarb, et al. states, “FCC practice, however, did not originate with professionals; this was not given to families; it was a response.”^{39(p92)} Family members first worked within their states at the grassroots level to improve healthcare service.³⁹ Over time, these individuals united into national organizations such as Family Voices, which was created in 1992 in collaboration with the Federal Division of Services for CSHCN.³⁹ Family-led organizations have worked to improve the lives of CYSHCN at the systems level, affecting services, policy, and program efforts. Family Voices’s statement on public policy and advocacy states that it works with governmental officials to improve health care for CYSHCN and their family members, advocate for family-centered care, and work to ensure that the family voice is heard.⁶³ Family Voices policy work focuses on several issues, such as insurance, SSI, support for family members of CYSHCN, and federal support for family-to-family groups and the Maternal and Child Health Block Grant program.⁶³

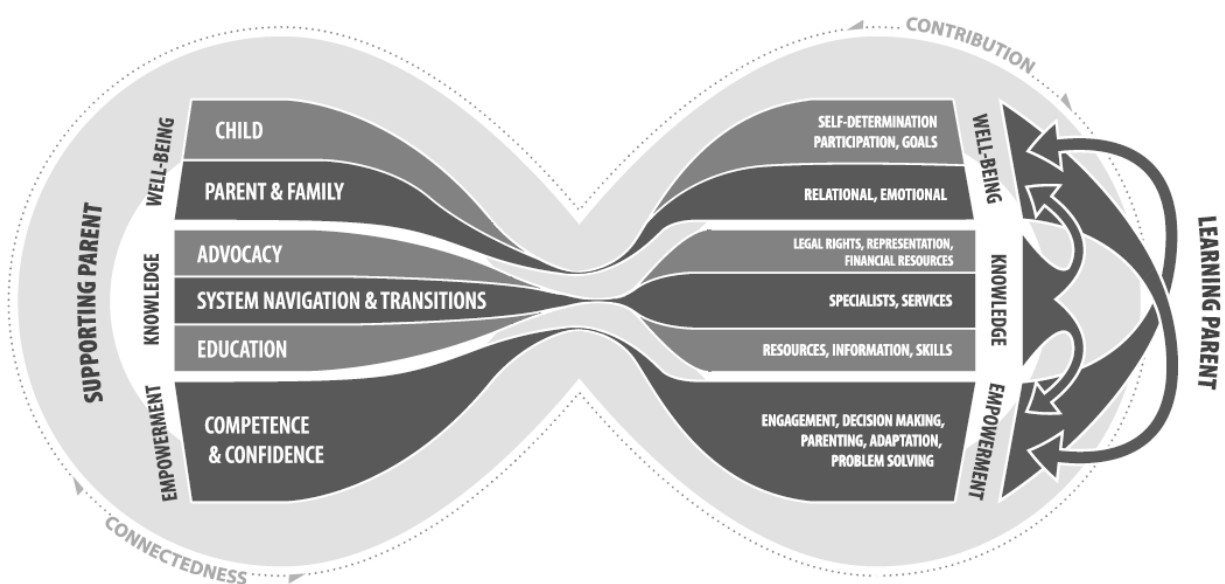
2.3.13 Benefits of Connecting with Family-Led Organizations

There are benefits for families of CYSHCN, and CYSHCN themselves, to being connected with family-led organizations at all levels and for multiple matters.^{22,33,35} Family-led organizations provide materials on disorders and prescriptions, and assistance with managing IEPs. Family-led organizations also provide training on understanding health insurance and communicating with doctors, among other services specific to each organization. Additionally, family-led organizations can provide one-on-one mentoring and emotional support.

When family members of CYSHCN, including CMC, are connected with parent mentors and family-led organizations, they benefit not only in improved navigation of community, education, and healthcare services, which improves access to these systems, but also in improved mental health.^{3,33} One-on-one emotional and social support reduces stress, anxiety, and feelings of isolation, as well as leads to enhanced involvement and access to care.³³

2.3.14 Final Conceptual Framework of Parent-to-Parent Support

A two-part study by Henderson, et al. (2014) and Henderson, et al. (2016) aimed to identify essential components of the parent-to-parent support connection using first a literature review to develop a conceptual framework,¹⁰ and then questioned 17-21 experts in the field in two stages to revise the conceptual framework.¹¹ This study focused on parents of children who were deaf or hard of hearing (D/HH).¹¹ The final conceptual framework identified three overarching themes of parent-to-parent support: well-being, knowledge, and empowerment, as show in the figure below.¹¹



© Henderson, Johnson & Moodie

Figure 2 Revised Conceptual Framework for Parent-to-Parent Support.

*Note: Order Detail ID: 71942060 American journal of audiology by American Speech-Language-Hearing Association Reproduced with permission of American Speech-Language-Hearing Association in the format Thesis/Dissertation via Copyright Clearance Center. Henderson, et al. (2016).¹¹

The helix shape represents both the supporting parent and the learning parent, as well as the information moving bi-directionally between them.¹¹ Connectedness and contribution both support this relationship.¹¹ The three overarching themes, well-being, knowledge, and empowerment are represented in a tiered fashion within the framework, and at the right, the arrows represent the interconnected nature among these three overarching themes for the learning parent.¹¹ Knowledge influences both well-being and empowerment, and well-being and empowerment influence each other.¹¹ Within each of the three overarching themes, components are shown for both the supporting parent and the learning parent.¹¹ Empowerment, the third tier, for example, encompasses competence and confidence in the supporting parent and several components in the learning parent, including engagement, decision making, parenting, adaptation, and problem solving.¹¹

Part one of this study concluded that parent-to-parent support is critical in the use of the medical home model for care of family members of children who are D/HH, as the supporting parents provide aid that healthcare providers cannot.¹⁰ Part two of the study proposed that the revised conceptual framework would have a functional role in the development of policy, programs, and FCC.¹¹ One participant in the second study stated, “We are constantly having to defend parent-to-parent support as an ‘add on’ to the journey as opposed to [an] ‘essential element’ so I think this model will give us the teeth to move parent-to-parent support into [a] systemic requirement.”^{11(p123)} The authors also acknowledged that this revised framework sits within its environmental context and that the challenges and the social determinants of health will have an effect on parent-to-parent support.¹¹

2.4 Studies of Parental Support from the Parent Perspective

In a paper on clinical issues in the *Journal of Obstetric, Gynecologic, and Neonatal Nursing*, one mother of two children with CF reflected on issues of having children with a genetic illness.⁶⁴ Gore Olsen shared her feelings of guilt over having a second child affected with CF, the stresses of family planning, the grief of not having a healthy child and of possibly not having other healthy children in the future, and the challenges of dealing with issues in the extended family when the child’s condition is genetic.⁶⁴ Gore Olsen went on to discuss the benefits of the parent-to-parent support received, including emotional and educational support, as well as a better understanding of why compliance with healthcare provider recommendations is so important.⁶⁴ At the time of publication, Gore Olsen was the executive director of the Indiana Parent Information Network, Inc.⁶⁴

Multiple studies have analyzed the support needs of, and the value of support for, parents of CYSHCN from the perspective of the parents themselves. One study by Pelentsov, et al. reported the results of an online survey taken by 301 parents of children with a rare disease living in Australia or New Zealand intended to inform best practices for developing supportive care for these parents covering six domains: 1) demographic details, 2) equity in care, 3) practical care needs, 4) relationships, 5) emotions, and 6) a summary.⁶⁵

Parents of children with rare diseases often face additional difficulties, including diagnostic odysseys, lack of nearby support groups, limited access to healthcare information and services, and the likelihood of feeling isolated.⁶⁵ In the second domain, approximately one third of parents reported having little to no support from government agencies, non-governmental agencies, and family or friends; 7% reported having no support from any of the above sources.⁶⁵ Within the third domain, the majority of parents reported needing more information about available services for their child (60.8-72.5%); and many parents reported being unable to access disease-specific support groups (42%), healthcare professionals who understood their child's diagnosis (37%), or psychological counseling (48%).⁶⁵

In regards to the fourth domain about their relationships, 58% of parents said having a child with a rare disease had reduced the number of friends they had.⁶⁵ A majority of parents (75%) reported that they had not met another family with a child like their own, 46% felt isolated, and 46% also "felt desperately lonely."^{65(p8)} In regards to the domain about emotions (the fifth), 37% of parents were being treated for depression, 41% for anxiety, and 10% for other mental health issues.⁶⁵ The article goes on to detail the strain for parents of children with rare diseases of maintaining friendships and the cyclical nature of the problem. It also states that friends and family may be intimidated by severity of the situation and not understand what the parents are going

through, and therefore distance themselves from the parents of children with rare diseases.⁶⁵ These findings bolster the idea that supporting parents have an opportunity to connect with learning parents in a unique way and make a significant positive impact in their lives. Supporting parents may offer encouragement and a sense of belonging that translates into better mental health for the learning parents and their improved ability to manage their child's healthcare needs.⁶⁵

Another study by Konrad that was designed to inform best practices for healthcare professionals reported the findings of interviews of 12 mothers of seriously ill, dying, or deceased children engaged through a palliative care program.¹³ The study unexpectedly found that parent-to-parent support was vital to the comfort of the parents.¹³ The parent-to-parent support described by the mothers included informal one-on-one interactions and formal engagement with P2P groups, illustrating the necessity of individualizing of parental support.¹³ Supporting parents, specifically only those with the lived experience of having a seriously ill or dying child, acted as mentors, confidantes, and consolers. The connection between supporting parents and learning parents helped these mothers cope, adapt, and identify positive aspects during their personal tragedy.¹³ Mothers were advised to be in the moment and appreciate the little things, they were encouraged to educate themselves about their child's illness, and they gained confidence in caring for their child.¹³ Mothers went from passive acceptance to becoming advocates for their child, and in so doing, became supporting parents for other mothers.¹³ This study not only illustrated the vital nature of parent-to-parent support, but also emphasized the need for "perceived sameness" for this connection to be authentic.¹³ Mothers also stated that they felt a connection with the healthcare workers with end-of-life training, likely because they had also achieved a version of this "perceived sameness."¹³ These healthcare workers were present during moments when other

healthcare professionals were not, and they had experienced these moments multiple times in oncology departments, hospice care settings, and neonatal intensive care units.¹³

The term “perceived sameness” was first referenced from self-help literature to apply to parent-to-parent support in a two-part, three year study conducting both qualitative and quantitative research done by Ainbinder, et al. and Singer, et al. on the value, impact, and meaning of parent-to-parent programs.^{1,2} Parents were recruited to participate in a study of a Parent-to-Parent self-help program using flyers in five states: Kansas, New Hampshire, North Carolina, South Carolina, and Vermont.¹ The parents had children with a range of disabilities from mild to severe and had diagnoses that included cerebral palsy, epilepsy, developmental delays, intellectual disabilities, learning disabilities, hearing and/or vision deficits, and chronic illness.¹ There were 24 learning parents paired with supporting parents interviewed in this study.¹

The interviews were analyzed using thematic analysis.¹ These interviews revealed that shared experiences between the learning and supporting parents promoted understanding, acceptance, and coping, and resulted in support that was bidirectional.¹ The supporting parents were seen as dependable and both normalized the learning parent’s experience and gave them hope for the future of their child. The supporting parents were described as a “reliable ally,” which the study found had four components: perceived sameness, comparable situations for learning relevant skills and gathering useful information, availability of support, and mutuality of support.¹ One aspect of the mutuality of support was that the relationship between the parents was equitable.¹ Many of the learning parents expressed satisfaction at the opportunity to provide support to their supporting parents.¹

The quantitative part of the study paired 56 learning parents with supporting parents (intervention group) and had a control group of 72 learning parents (not in crisis) who were placed

on a waitlist to be paired with a supporting parent.² All parents completed pre- and post-tests designed to measure acceptance of family and disability, perceptions of coping efficacy, empowerment, and progress in meeting the primary need for which they contacted Parent-to-Parent.² The first was measured using the Source of Strength and Family Closeness subscale of the Kansas Inventory of Parental Perceptions (KIPP), which found a significant improvement in attitudes related to cognitive adaptation in those in the intervention group.²

Perception of coping efficacy was measured using the Parent Coping Efficacy Scale (PCES), which was developed during this study, and had two subsections: Coping with Child and Coping with Family.² A statistically significant difference between those in the intervention group and those in the control group was seen only in those who scored below 3.07 (out of 5.0) on their pre-test, which means that only those with lower initial perceived coping efficacy improved significantly in the intervention group.² Empowerment, as measured by The Family Empowerment scale, was not found to be significantly different between those in the intervention and control groups.² Progress in meeting the primary need identified by parents as the major reason they contacted P2P was measured with one question: “How much progress have you made in meeting this need?”^{2(p225)} A significant increase in measured progress was found in the intervention group when compared to the control group.²

Finally, perceived helpfulness of the program was reported in 89% of the individuals in the intervention subset.² This study quantified specific benefits of parent-to-parent support. The data suggested that parents (who were not in crisis) received multiple benefits from participating in the Parent-to-Parent program and being paired with a supporting parent, including increasing feelings of being able to cope, having a more positive perspective, as well as supporting other parents.² This study provided the first quantitative evidence on the efficacy of parent-to-parent support.²

One previous study by Ireys, et al. attempted to provide quantitative evidence for the outcomes of family-to-family support, and while the data were internally consistent, methodological issues prevented the study from reaching statistically significant results.⁶⁶ This three-part study of randomized trials evaluated populations with different conditions, including 1) a wide range of conditions, 2) arthritis, or 3) a combination of diabetes, CF, sickle cell anemia, and moderate to severe asthma, respectively.⁶⁶ Studies 2 (n = 53) and 3 (n = 193) matched diagnoses of the children of the supporting and learning parents, where Study 1 (n = 365) did not.⁶⁶ The support programs were intended to provide informational, affirmational, and emotional support in order to affect parental mental health.⁶⁶ Supporting parents were tasked with calling their assigned learning parent every two weeks, meeting with them six times during the study, and participating in three special events for all families.⁶⁶ Families in the control group were given a phone number of another experienced but untrained mother.⁶⁷ Pre- and post-tests (at 12 months) were administered to mothers to measure anxiety levels using the subscale of the Psychiatric Symptom Index.⁶⁶ All three studies showed a general decrease in anxiety among high stress participants in the experimental group when compared to the control group.⁶⁶ Suggestions for future studies included incorporating qualitative analysis to further clarify results.⁶⁶

A study by Kerr, et al. conducted in Scotland utilizing qualitative analysis investigated the impact of parent-to-parent support on parents who had a child with a congenital limb deficiency.¹² Both couples (34) and mothers (29) were interviewed and the transcripts were analyzed using thematic analysis.¹² While the conclusions in this paper were consistent with the previously described studies, the authors commented more specifically on the need for parent-to-parent support as opposed to support from healthcare professionals, the community, or family and friends.¹² In addition to healthcare professionals having little experience with congenital limb

deficiencies, and mothers feeling uncomfortable taking their baby out because they felt their child was very different, family and friends were usually not able to provide comforting support. As stated in the paper, friends and family were going through their own grief process and as such placed additional strain on the situation in spite of their efforts.¹²

Parents of infants with structural birth defects registered in the Utah Birth Defect Network were invited to be a part of a study by Mathiesen, et al. where they participated in focus groups or interviews to determine parental needs, perspectives on support groups, and recommendations for a parent-to-parent support network.¹⁴ Qualitative analysis was performed on the recordings of the four focus groups, including a total of 24 parents, and seven individual interviews.¹⁴ Several parents reported the inability to locate parent-to-parent support consistent with the anomalies of their child and/or in nearby locations.¹⁴ When asked if they would have found a parent-to-parent network useful, 25 of 31 participants agreed, while most of the six participants who did not agree could still see the benefit for different situations and/or other individuals.¹⁴ While participants had differing personal preferences regarding parent-to-parent support groups, they ultimately recommended a flexible program to meet parental needs.¹⁴ The authors concluded that there was consistent desire and need for statewide parent support groups to be associated with birth defect registries and present in all hospitals to care for parents of children with structural birth defects.¹⁴ In lieu of this network, the authors emphasized the need for healthcare professionals to both have knowledge of and inform parents of existing parent-to-parent support groups.¹⁴

2.5 Studies of Parental Support from the Leader Perspective

Not many articles report on family-led organizations from the perspective of the leaders of these organizations. Two articles reported the results of a survey of directors of genetic organizations that provide support groups for individuals with genetic disorders.^{15,16} The focus of these articles was to describe the services provided by, and the importance of, these organizations.^{15,16} The first article by Black, et al. reported data collected from a survey of the directors of genetic support groups that focused on details of the service organization.¹⁶ A second survey was administered to the members of specific support organizations that focused on their connection to the organization and their satisfaction regarding acquiring genetic information.¹⁶ The directors of these organizations (n = 76) reported that education of both the public and healthcare professionals was their main service provided.¹⁶ The members of these organizations (n = 931) indicated that they have experienced barriers to obtaining information related to finding knowledgeable professionals, travel time to specialists, and costs.¹⁶ They also reported difficulties in obtaining related services.¹⁶ The authors suggested better communication and improved referrals among professionals, and an increase in genetics education to the public.¹⁶

The second article by Weiss also reported on this survey of directors of genetic service organizations, elaborating on the need for specialists and their ability to refer to these support groups.¹⁵ Additionally, this article provided guidelines for healthcare professionals who are interested in helping patient leaders organize the first meeting of a new support group and/or being a consultant to a support group.¹⁵ These articles conclude by stating the importance of integrating support groups into the treatment of those with genetic disorders.^{15,16}

Early Hearing Detection and Intervention (EHDI) is a standardized program for identifying infants with hearing loss.¹⁰ This process begins with screening newborns for hearing loss by one

month of age.⁶⁸ If indicated, this is followed up with hearing tests and a diagnosis by three months of age, and intervention services by six months of age.⁶⁸ Parental support is integral to the success of the EHDI program and subsequent development of language by the child who is D/HH.¹⁹ While accessing both assistive technology and specialized healthcare professionals is recognized as important, communication and language development are also affected by interaction and bonding between the child and parents, parental success in being emotionally available and coping with grief and stress, as well as the child's self-development.¹⁹

One study by Bradham, et al. surveyed 47 state coordinators of EHDI programs regarding family support in an effort to improve program effectiveness and infant outcomes.¹⁹ Analysis of the surveys identified several strategies for improvement, including developing parent support groups if not already in place and having parents of children who are D/HH work as paid staff at these support groups.¹⁹ Those EDHI state coordinators already working with formal family support organizations reported significant benefits from the collaboration.¹⁹ Family-to-family support not only benefits the parents, but also the child in the family, and can improve follow-up outcomes.¹⁹ Bradham, et al. states, "Improving the well-being of the family as a unit provides a favorable environment to support the child's developmental and communicative success—a goal that remains at the center of all EHDI programs."^{19(p193)}

Lastly, one study by Behl, et al. surveyed directors of family-led organizations directly to determine how their organizations support families with hearing-related concerns and to identify areas for improvement.¹⁷ The state-wide family-led organizations that were contacted from across the country included F2F groups, SAOs, P2P groups, and PTI Centers, and resulted in a total of 104 survey responses.¹⁷ In addition to previously mentioned information that family-led organizations provide, information on legal services was reported as provided by 67% of

respondents.¹⁷ In addition to their many other activities previously discussed, respondents also reported working on grants with state EHDI programs (11%).¹⁷

Given the choice to indicate challenges (in the form of checking all that apply from a given list), respondents listed knowing about D/HH financial resources (62%), having materials available in other languages (47%), and engaging D/HH families (44%) as challenges they faced.¹⁷ When given the opportunity to write in challenges that they faced, participants reported previously described access issues, and also reported that locating support for CYSHCN with multiple diagnoses was also an issue.¹⁷ One area identified for improvement was the small percentage of families of children who are D/HH that contact these family-led organizations (less than 6%), which corresponds with parents reporting that they lack information, access to resources, and family support.¹⁷ Increased awareness of, and referrals to, these family-led organizations may close this divide.¹⁷

2.6 Grief and Mourning

The response to both the loss of having a “normal” child and (possibly) the loss of having additional children in the future is complex and involves multiple psychological processes. These processes include coping strategies, defense mechanisms, and grief reactions. Guilt may also play a part in these psychological reactions.⁶⁹

It has been reported by Weil that a negative experience can destroy a person’s belief in an orderly world.⁶⁹ After a negative, unexpected event occurs and changes an individual’s perception of the world, feelings of powerlessness are not uncommon.⁶⁹ These may also be coupled with feelings of meaninglessness.⁶⁹ Thus, an individual may need to search for new meaning in which

the negative event can be explained.⁶⁹ This sometimes manifests in the search for a cause of the event, for which a person may take personal responsibility and feel guilt, which is discussed below.⁶⁹

Coping strategies during this process may be problem focused, or emotion focused, and while they are not mutually exclusive, emotion focused coping strategies are more common in situations that are unchangeable.⁶⁹ An additional coping strategy is that of reappraisal, where the individual has a more positive outlook and wider perspective on their situation.⁶⁹ The use and type of coping strategies may change as the individual's or family's situation changes.⁶⁹ These coping strategies are associated with defense mechanisms, which are discussed below, but individuals are more conscious of coping strategies than they are of defense mechanisms.⁶⁹

Defense mechanisms are unconscious methods for our psyche to endure a negative experience.⁶⁹ Some of these defense mechanisms include repression, or the lack of conscious awareness of certain feelings or experiences; displacement, where directed emotions are shifted from the cause of the emotions to a less anxiety-producing outlet; reaction formation, when an individual expresses an emotion the opposite of what they are actually feeling; and projection, where certain emotions are perceived as coming from another individual.⁶⁹ They may also include intellectualization, which is less emotional and more rational, or sublimation, which is dealing with difficult emotions by turning them into socially acceptable outlets.⁶⁹

Grief reactions have been described as stages and are constructs used to describe phases in the mourning process, which is individualized and non-linear. One longitudinal cohort study by Maciejewski, et al. did support the stage theory of grief with quantifiable data,⁷⁰ but these stages are labels used to discuss this process. An original stage theory of grief proposed by Bowlby and Parkes included four stages: shock-numbness, yearning-searching, disorganization-disrepair, and

reorganization.⁷⁰ The updated five stages in the Kubler-Ross model include denial, bargaining, anger, depression, and acceptance.⁷¹ Shock is also stated as an initial reaction.⁶⁹

True denial, where there is a continuation of the defense mechanism of repression, is rare.⁷² The stage of denial can also be referred to as a stage of disbelief or deferral.⁷² Disbelief is characterized by confusion and has its origins in dissonance regarding the information given, whereas deferral is characterized by avoidance of implications and has its origins in the lack of physical or psychological resources.⁷² Another mimic of denial is dismissal, which is characterized by a denial of the professional's legitimacy and has its origins in feelings of entrapment or betrayal.⁷²

Bargaining may be a hope for a miraculous solution, where atonement is offered by the individual experiencing the grief.⁶⁹ The stage of bargaining is sometimes replaced with the stage of guilt, which is discussed below. Anger at the situation is common and may be dealt with unconsciously with a variety of defense mechanisms.⁶⁹ Depression may result directly from the loss experienced or from the individual's reaction to the loss (guilt, for example),⁶⁹ and may last for months afterwards.⁷⁰ Lastly, the acceptance stage is more accurately referred to as the adjustment to the "new normal."

An updated version of the five stages of grief is the Kubler-Ross Change Curve, which includes seven stages: shock, denial, frustration, depression, experiment (or beginning to engage with a new situation), decision (or learning to function and be more positive in the new situation), and integration (or integrating changes to the new situation).⁷³

Guilt is a common, natural response to a negatively perceived event that occurred, which was out of a person's control.⁷⁴ Feelings of guilt may increase when the cause of the event is unknown.⁷⁴ Kessler, et al. stated that, "guilt is generally associated with responses of self-reproach

to violation of internal standards”^{74(p675)} and may lead to a negative self-image.⁷⁴ It has also been reported by Kessler, et al. that guilt (or personal responsibility) can serve as a defense against powerlessness.⁷⁴ As personal responsibility suggests control over a situation, guilt may defend the psyche from the greater feelings of loss and a shattered belief in a caring world.⁶⁹

Family members of CYSHCN often feel more comfortable discussing their situation and grieving process without fear of judgement with another family member of a CYSHCN.⁶⁹ A family support network provides a space to confide, cope, learn, and adapt.⁷⁵ A supporting parent can provide the opportunity to develop necessary skills, and suggestions for navigating systems and services, and proof that there can be survival through grief.⁷⁵ And learning parents can gain a sense of normalization from the supporting parent, and hope from seeing older CYSHCN.⁷⁵

2.7 Qualitative Research

While there is no universally agreed upon definition of qualitative research, there are several descriptions that are helpful in conceptualizing qualitative research. One description cited by Beeson states that it is “research in which the investigator attempts to study naturally occurring phenomena in all their complexity”^{76(p22)} and another states it is “multimethod in focus, involving an interpretative, naturalistic approach to its subject matter.”^{76(p22)} Qualitative research is used in education, psychology, public health, anthropology, and sociology, among other disciplines.

Another description of qualitative research cited by Beeson states that “the strength of qualitative research is precisely its respect for the empirical world, that is, its potential for yielding verifiable knowledge of human group life and human conduct. ... The term ‘empirical’ refers to knowledge gained from observation.”^{76(p26)} One tenet of qualitative research is that there is no one

right perception of reality, which includes the researcher's own perception of reality. This requires that the researcher attempt to be as objective as possible, all while recognizing how their own subjectivity will influence the research.⁷⁶ Another principle of qualitative analysis is that "theory both guides qualitative research and results from it. ... In [some] study designs, researchers avoid imposing a theoretical framework, letting theory emerge from the data analysis."^{77(p141)} The most common form of data collection for qualitative analysis is that of interviews, which are then transcribed.⁷⁶

Thematic analysis is one type of qualitative analysis and has several stages: familiarizing yourself with the data, generating codes, searching for themes and subthemes, reviewing themes (which may include making a thematic map), and defining and interpreting themes.^{77,78} This process is non-linear and iterative as needed.⁷⁸

One approach to analyzing qualitative data is that of thematic analysis, which is described by Braun, et al. as "a method for identifying, analyzing, and reporting patterns (themes) within data. It minimally organizes and describes your data set in (rich) detail. However, frequently it goes further than this, and interprets various aspects of the research topic."^{78(p79)} Thematic analysis is one useful approach to elucidate information about participation in an organization, and to acquire different perspectives at multiple levels.⁷⁶

Several procedural decisions complement each other when completing thematic analysis. Data analysis can be done using an inductive coding method, or 'bottom up,' approach, which is data driven and allows for coding and analysis to be performed without adherence to a specific theory.⁷⁸ Identified semantic themes seek to describe the surface meanings and form of the data.⁷⁸ When all interviews are combined into one whole data set and analyzed together a rich description of the data is generated.⁷⁸ This technique is applicable to areas of research that are not heavily

studied and populations that are not represented in the literature.⁷⁸ Lastly, a realist method can be used, which conveys reality according to the participants, as opposed to conveying reality from the broader social context.⁷⁸ These approaches are generally used together.⁷⁸

In contrast, a deductive coding method, or “top-down” approach, which focuses on a specific theory, can be used.⁷⁸ Identifying latent themes, or underlying themes interpreted from the data, is generally paired with analyzing the data, or a subset of the data, for a specific topic, such as a latent theme.⁷⁸ This focused, in-depth approach is usually employed with a constructionist method, which conveys realities as influenced by social context.⁷⁸ However, the use of these methods can be interchanged to create combinations beyond these two categories.⁷⁸

As this study looks to perform data analysis without basing it on a previously described theory, an inductive coding method was chosen. The analysis of the entire data set was performed, semantic themes were identified, and a realist method was used. This combination of methods can be used to provide a broad, rich description of a qualitative data set, enabling an initial exploration of a topic and population that are scarce in the current literature.

2.8 Purpose of Study

The structure and context of family-led organizations and their partner organizations, as well as the political and financial milestones that have fostered these organizations have been discussed in the prior sections. While these organizations have evolved and continue to help individuals access healthcare and other supports, there are still many widespread barriers to access. These barriers include lack of care coordination, financial difficulties, geographical and cultural impediments, health insurance problems, and racial, and ethnic disparities. Studies have been

performed analyzing these issues from the perspective of family members of CYSHCN, and some leaders of support groups, but there is a lack of literature from the perspective of leaders in family-led organizations.

In this study, the leaders of family-led organizations within the NYMAC region have been interviewed directly, increasing the scope of this area of research and gaining new insight from the perspective of these leaders. The recorded interviews were analyzed using thematic analysis to elucidate themes throughout the data set. While family-led organizations provide assistance on the individual level and work to advocate for change at the systems level, integrating these organizations into systems of care has the potential to greatly reduce the previously described barriers to care. Collaboration across organizations on efforts to improve access to services could make substantial positive change and including leaders of family-led organizations into future community-engaged research projects would be beneficial to the outcome of the research. Additionally, supporting the widespread adoption of the medical home model has considerable potential to improve healthcare. The results of the study may help to identify areas of future growth, research, and collaboration.

3.0 Manuscript

3.1 Background

Caregivers of children and youth with special health care needs face not only the multifaceted challenges of accessing quality care, but also the demands of meeting the child's or youth's needs for special health care.^{1,2,50} Children and youths are considered CYSHCN if they are at risk for a chronic condition and require care beyond what would be expected for a child or youth.⁹ CYSHCN may require additional behavioral, mental health, physical health, educational, and/or community services.⁹ The 2005-2006 NS-CSHCN data were the subject of a secondary quantitative analysis performed by Kuo to determine the care requirements of children with medical complexity (CMC), a more complex subpopulation of CSHCN.⁶ The survey is designed to provide approximate numbers of CSHCN, including CMC, within each state and describe the healthcare use and burden of this population.⁶ The secondary analysis findings, which covered the degree of success in accessing particular services, reported widespread difficulty in systems navigation and the importance of care coordination for CMC.⁶ Poor or absent care coordination was found by Beene-Harris, et al. to be an institutional barrier to access for genetic services, specifically.⁵ This barrier included aspects such as access to information, follow-up care, and referral to support groups.⁵

Family-led organizations provide important support to family members of CYSHCN and CYSHCN themselves.^{1,2} All staff members of these organizations are family members of CYSHCN, including parents and siblings.³ The leaders of family-led organizations have those positions because of their personal experience, parental perspective, and their leadership abilities.

There are several different types of these family-led organizations, each with its own specialty; Parent-to-Parent (P2P) groups work to match family members of CYSHCN one-on-one with other supporting family members of CYSHCN.³ Family-to-Family (F2F) groups focus on helping families with healthcare issues.³ State Affiliate Organizations (SAOs) work to progress the mission of the national Family Voices organization,³ which is to keep the family voice at the center of healthcare.⁴ In some states, combinations of family-led organizations are housed at the same location. Table 4 below shows the structure of P2Ps, F2Fs, and SAOs within an eight state region (including District of Columbia).

Table 4 Family-Led Organizations (P2P, F2F, & SAO only) in Region, adapted.

| State | Subset of Family-Led Organizations in the NYMAC Region | | |
|----------------------|--|--|--|
| | P2P | F2F | SAO |
| Delaware | Delaware Family Voices | | |
| Maryland | Parent to Parent of Maryland at the ARC MD | Parents' Place of Maryland, Inc. | |
| New Jersey | Statewide Parent Advocacy Network (SPAN) | | |
| New York | Parent to Parent of NYS | | |
| Pennsylvania | Parent to Parent of PA | Parent Education and Advocacy Leadership Center (PEAL) | |
| Virginia | Center for Family Involvement | Family to Family Network of Virginia - Center for Family Involvement | |
| West Virginia | | West Virginia University's Center for Excellence in Disabilities | West Virginia Parent Training and Information |
| District of Columbia | | Advocates for Justice and Education | Family Voices of the District of Columbia Inc. |

*Note: Adapted from Referral to Support and Partnering with Family-Led Organizations.³

One group working to bring together leaders of family-led organizations, healthcare professionals, and service delivery specialists is the Regional Genetics Networks and their National Coordinating Center (NCC). The New York Mid-Atlantic Consortium (NYMAC)

Regional Genetics Network (RGN) is one of several RGNs that partner with family-led organizations to improve access to genetic services, specifically focusing on underserved populations.²¹ The NYMAC region includes Delaware, Maryland, New Jersey, New York, Pennsylvania, Virginia, West Virginia, and the District of Columbia. The family-led organizations and RGNs also work together to train families for leadership roles and empower families to become involved in promoting change at multiple levels.

Family-led organizations not only work to directly help family members of CYSHCN and CYSHCN themselves, but also work to advocate for family-centered care, which emphasizes partnership between the family and the health professional.⁷ Family-centered care is an integral part of the medical home model endorsed by the American Academy of Pediatrics.⁸ The medical home model emphasizes continuous, family-centered, coordinated care.⁸ Care coordination is the coordinated effort of connecting patients and their families with services to ensure quality healthcare.¹⁸ The medical home model, including care coordination, has the potential to minimize barriers to access to care by locating healthcare professionals, improving communication, reducing system fragmentation, minimizing cultural barriers, and removing the responsibility of care coordination from the family members of CYSHCN.^{8,33,35} Family-led organizations can benefit family members of CYSHCN and CYSHCN themselves across multiple systems, including supporting their emotional needs, assisting with educational issues, providing information on specific disorders, and providing training on health insurance, among other topics.

The conceptual framework of the parent-to-parent connection, in relation to parents of children who were deaf or hard of hearing (D/HH), was broken down into several overarching themes in one study.^{10,11} This two-part study by Henderson, et al. (2014) and Henderson, et al.

(2016) found that well-being, knowledge, and empowerment were shared in the unique relationship offered by parent-to-parent support.^{10,11}

Family-led organizations produce positive effects by supporting parents; this support has been documented mainly from the perspective of the parent. The first study to report statistically significant benefits of P2P support both quantitatively and qualitatively analyzed empirical data from a longitudinal two-part study done by Ainbinder, et al. and Singer, et al.^{1,2} This study detailed specific benefits of parent-to-parent support and provided evidence beyond anecdotal information on the efficacy of this support.^{1,2} Other studies later performed analysis of interviews with parents of children with a congenital upper limb deficiency (Kerr, et al.),¹² and parents of seriously ill and dying children (Konrad),¹³ and found that parent-to-parent support was considered helpful. One study done by Mathiesen, et al. interviewed and conducted focus groups with parents of children registered with a structural birth defect; after these discussions, the authors suggested the association of a parent support group with both the birth defects registry and the hospital system.¹⁴

Two articles by Black, et al. and Weiss reported the results of a survey of directors of genetic support groups,¹⁶ and genetic organizations that offered services to families¹⁵ to describe the services provided by, and importance of, these organizations. The directors of these organizations (n = 76) reported that education of both the public and healthcare professionals was their main service provided.¹⁶ These articles conclude by stating the importance of integrating support groups into the treatment of those with genetic disorders.^{15,16} Early Hearing Detection and Intervention (EDHI) state coordinators were surveyed in a study performed by Bradham, et al., and those already working with formal family support organizations reported significant benefits from the collaboration.¹⁹ Family-to-family support not only benefits the parents, but also the child in the family, and can improve follow-up outcomes.¹⁹ A survey of leaders of family-led

organizations was also done by Behl, et al. and in addition to previously mentioned information that family-led organizations provide, information on legal services was reported as provided by 67% of respondents.¹⁷ Respondents also reported working on grants with EHDI programs (11%).¹⁷

The current study involved interviewing leaders within family-led organizations in the NYMAC region, which includes Delaware, Maryland, New Jersey, New York, Pennsylvania, Virginia, West Virginia, and the District of Columbia. By interviewing leaders of family-led organizations, this study captures the unique perspective of those who are both leaders and family members of CYSHCN. These leaders represented themselves directly, providing new insight and increasing the scope of this area of research. These interviews covered topics including working with providers, care coordination, and issues of access. The results of this study may be applicable to healthcare providers, including those working in mental health; members of the education system; county, community, and other service providers; governmental organizations; and organizations working in this field, including the family-led organizations themselves.

3.2 Methods

3.2.1 Study Design

The study consisted of conducting interviews with leaders of family-led organizations. One-on-one interviews of participants allowed for the confidential discussion of topics outside the presence of their colleagues. The study (STUDY19050146) was reviewed and approved by the Institutional Review Board at the University of Pittsburgh and the approval letter is shown in **Appendix A**.

3.2.2 Participant Recruitment

Leaders were invited to participate via email contact (**Appendix C** contains the email invitation). The email invitation contained information describing the lead researcher, what documents the data would be used in, the criteria for being a participant, the details of the data collection and analysis process, and the goals of the project, and also asked individuals to respond if interested in participating. Leaders of family-led organizations were also invited to participate during a conference call between personnel from NYMAC and participants from the network of family-led organizations in that region. Only leaders who emailed responses expressing interest in participating in the study were invited to participate. Consent was obtained throughout the study, and formal consent using the consent script (**Appendix D**) was finalized after the interviews.

3.2.3 Study Participants

Study participants are leaders of family-led organizations in the NYMAC region, which includes Delaware, Maryland, New Jersey, New York, Pennsylvania, Virginia, West Virginia, and the District of Columbia. The family-led organizations were identified by the Genetics Education and Family Support Center,³ which is led by the Genetic Alliance. Leaders of family-led organizations are required to be family members of CYSHCN. A total of six leaders within family-led organizations were interviewed via Skype for Business, representing five states and multiple levels within the organizations.

3.2.4 Data Analysis

This study used thematic analysis to analyze semi-structured interviews of leaders in family-led organizations within the NYMAC region. Interview questions went through multiple iterations and were designed to be open ended, broad in scope, and absent of leading questions. The interview guide is shown in **Appendix B**. Thematic analysis is well suited to analyzing participant perceptions across multiple levels of an organization.⁷⁶ Specifically, an inductive coding method was used. This method is data driven and does not use a previously described theory to generate codes or conduct analysis.⁷⁸ Themes identified were semantic, or those seeking to describe the surface meanings and form of the data.⁷⁸ All interviews were combined into a one whole data set and analyzed together. This technique generates a rich description of the data, applicable to areas of research that are not heavily studied and populations who are not represented in the literature.⁷⁸ Lastly, a realist method was used, which conveys the realities of the participants, as opposed to the broader social context.⁷⁸

Notes were taken during the recorded interviews and after interviews were completed, they were transcribed verbatim by the researcher. Transcripts were analyzed using thematic analysis, which has several stages: familiarizing yourself with the data, generating codes, searching for themes and subthemes, reviewing themes (including making a thematic map), and defining and interpreting themes.^{77,78} To become familiar with the data, each transcript was read and notes were taken on specific excerpts, coding ideas, and overarching themes. Preliminary codes were generated when re-reading the transcripts; these codes were then edited by both breaking down a code into multiple codes and by combining multiple codes into one code as deemed appropriate. The transcripts were coded manually using the updated code book and then the coding was refined as the transcripts were coded again in Microsoft Word.

Preliminary thematic maps were generated by organizing codes into themes and relating themes to one another. The coded transcripts were reviewed one last time in Microsoft Word to generate the final coded transcriptions for the entire data set, and then sorted by code. All transcripts were combined into one file and again sorted by code. A final thematic map, which also identified which codes were associated with which themes, was made. Files were then generated using Microsoft Word for each theme and subtheme identified by incorporating the excerpts with the assigned codes from the entire data set.

3.3 Results

A total of 17 individuals were contacted directly with an invitation to participate in the study, and two individuals were recruited by word of mouth. Of the seven individuals who indicated interest in participating in the study, one could not participate as the data collection timeframe had passed. A total of six individuals were interviewed, representing five different states in the NYMAC region. All six participants were female, and all were a family member of a CYSHCN. Unique identifiers are not used for identifying the participant quotes below in the interest of keeping the participants from being identified and protecting participant privacy and confidentiality. The response rate of those contacted directly was 29%. The total response rate including all interested individuals was 37%.

Several themes and subthemes were identified from the analysis of the interviews with leaders of parent-led organizations. A table of these is shown below.

Table 5 Themes and Subthemes Identified through Analysis of Interviews.

| Theme | Subtheme(s) |
|--|---|
| Experiences of Being a Family Member of a CYSHCN | Difficult Emotions Associated with the Experience |
| Working with Providers | Expectations and Hopes for CYSHCN |
| | Care Coordination |
| | Experiences with Genetics |
| Being a Leader in a Family-Led Organization | Issues of Access |
| | Progression of Family-Led Organizations |
| | Ideological Differences Among Leaders of Family-Led Organizations |

The themes include I) Experiences of Being a Family Member of a CYSHCN, II) Working with Providers, and III) Being a Leader in a Family-Led Organization. There were seven subthemes that were related to one of these three themes. One subtheme, Difficult Emotions Associated with the Experience, connected solely to being a family member of a CYSHCN. Many family members of CYSHCN discussed the psychosocial challenges they faced, and continue to face, over time. These challenges are included within this theme.

Another theme, Working with Providers, connected to both themes of being a family member of a CYSHCN and being a leader of a family-led organization. Participants had many impactful experiences working with providers as both family members of CYSHCN and as leaders in family-led organizations, both of which are included in this theme. Three subthemes related to the theme of working with providers include: 1) Expectations and Hopes for CYSHCN, 2) Care Coordination, and 3) Experiences with Genetics. There were many components to the participants' discussion of working with providers, which generated these three subthemes.

Two subthemes, Issues of Access and Progression of the Family-Led Organizations were connected solely to the theme of being a leader of a family-led organization. Both of these

subthemes were discussed mainly from the perspective of a leader of one of these organizations. One subtheme emerged from the subtheme of the progression of family-led organizations, which was Ideological Differences Among Leaders of Family-Led Organizations. This subtheme emerged directly from the subtheme of the progression of the organizations and was very specifically discussed from the perspective of a leader of one of these organizations. These themes and subthemes are described below. A final thematic map of the themes and subthemes identified is shown in Figure 3.

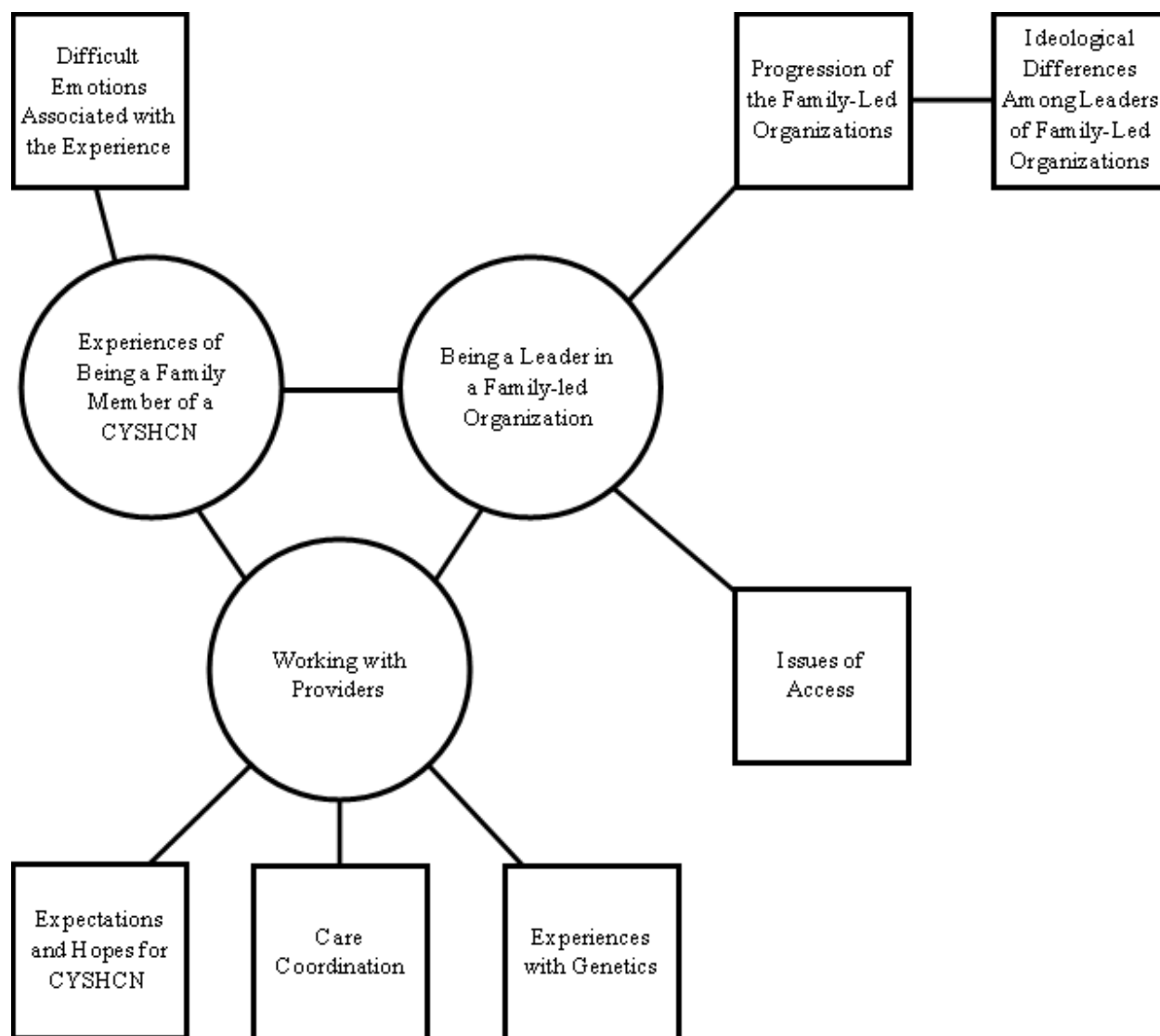


Figure 3 Final Thematic Map.

3.3.1 Experiences of Being a Family Member of a CYSHCN

While each participant already had their own set of skills, education, and life plans, the beginning of their journey to becoming a leader of a family-led organization began when each became the family member of a CYSHCN. Instantaneously, these individuals started at the beginning of a steep learning curve, one which would include challenges and emotional ups and

downs. Several participants described the conditions they found themselves in at this time, including being in survival mode, as well as the transition process they had to go through. One participant shared:

How do you bond with a ... child that you don't get to see? Because, to be honest, my husband and I would go and visit [her], we would see her during the day, in between work hours, we'd go back at night and see her, and then we were like, ok I guess we can go to the bar ... visiting hours are over, it's nine o'clock. And so, we acted like typical young people, we were like we'll just go have a beer, and people were like aren't you new parents? And we were like, yeah, kind of, but she doesn't live with us ... and so ... there was a disruption in some of that and it was a really weird life that first year of, we are parents, but we're not, because we didn't feel like we were.

Many participants shared advice they received from other family members of CYSHCN and support organizations. The same participant went on to describe connections that helped the family persist:

We relied on the nurses to help teach us things; I relied on my own kind of upbringing, and really, it was that connection to another parent, and getting another parent who really kind of, helped kick my ass into moving forward, and say, there's more out there than just what the professionals are telling me, and so I think for us it was a lot of ... it was kind of a combination of all of that.

Family-led organizations as well as other support organizations were mentioned as helpful by multiple participants. One participant described getting connected with another support organization and its impact on her perspective:

Initially, I was just learning and finding out things, and the other organization I got connected with was National Family Caregivers, it's now known as the Caregiver Community Action Network ... and one of the things I found out was, caregiving is a lifespan issue, it's not just an elder care issue. I was sitting there thinking, yeah, not all moms that ... have [a] kid that's four and five are diapering them or giving them baby food, like this lightbulb went off.

These caregivers of CYSHCN have faced fundamental challenges across the lifespan of the child or youth. Some of these were difficulties working within the healthcare system and barriers to access to rights, which included education. These participants each became the child's

or youth's dedicated advocate in all aspects of their life. They described multiple instances of making critical decisions regarding the lifepath of the child or youth. One participant talked specifically about this decision-making process:

Well, as a parent again, information is power. You need to know what you're dealing with to know what your choices are. One of the best things that somebody ever told me [is] ... as a caregiver, you may not have all the choices that you had before, and you may not like some of the choices that you have, but you still have the power to choose. And I think that was so profound, because in our case, it was, ok do we want [one type of treatment or another]... And, it's like yeah, we don't like those choices, but we still have some control over what happens.

Several participants talked about the decision to not send their CYSHCN to a segregated special education program, or a group home. One participant shared:

A lot of it was just sheer determination as a parent that I wasn't going to have [her] be in a group home and be in a sheltered workshop where she made ten cents an hour. That just was not, that just wasn't going to happen. I wasn't raised that way; I was around people with disabilities. [She] belonged, needed to be included in her school and in her community and she needed to have some pride and self-worth.

Several individuals described the efforts to ensure care for the CYSHCN. Each CYSHCN who was discussed had different needs and strengths that affected the details of their lives. Each family member of a CYSHCN devoted substantial amounts of time, resources, and energy into supporting that child or youth in a variety of conditions. The participants shared how these CYSHCN had succeeded in multiple ways. One participant shared:

My daughter graduated from [college] ... and I was extremely proud of her ... from going to having all of these health issues, and having problems with reading and everything, and then she graduated from college and is doing really good. I'm super proud of her for that, and you don't ever want to set a bar too high, but you want to set that bar Don't ever let them be like, I'm dyslexic so I'm not ever going to be good at that; no, you can, with work, you can, with practice, you can. We are all not perfect, we all make mistakes, we all constantly learn things every single day.

3.3.1.1 Difficult Emotions Associated with the Experience

While the participants described how they devoted themselves to the care of their own family member(s), as well as working in their professional lives to better the lives of other family members of CYSHCN and CYSHCN themselves, this did not preclude their having difficult emotions associated with certain aspects of their experiences. These difficult emotions generally did not necessarily occur in mutually exclusive stages one after the other. These emotions may have been concurrent with, or subsequent to each other, as well as cyclical over time. Additionally, individuals may have sometimes felt conflicting emotions at the same time, such as joy over having a child and also sadness that the child was experiencing adversity. Several different emotional responses described by the participants, such as loss, guilt, regret, desperation, powerlessness, and righteous anger are detailed below, each in its own section.

Loss

After having an affected child, or receiving a diagnosis, family members of CYSHCN may feel grief associated with the loss of the dream of the family member they thought they would have or (possibly) with the loss of the dream of having additional children in the future. Several participants talked about the multiple instances of loss they experienced and the subsequent processes of mourning. One participant shared:

I guess it's that hopes and dreams piece that we all lose or thought we lost when we get a diagnosis for our kids and imagine what we think their life could and should be and then we find that they are diagnosed with something.

A second participant shared:

When she was much younger, we were making decisions as to whether or not to have other children and initially we only knew about the [one] disease, but they decided that that was genetic. And, interestingly enough I told both my brother and my sister, please check when you're ready to have kids, have level II diagnostic ultrasounds ... and both of my brother's kids had the [anomaly]. ... And then we were still like, well maybe ... and then when she

was seven, we got the [second] diagnosis, and we're like nope, nope. Again, they thought it was genetic. It turns out that my nephew on my husband's side also has [that diagnosis], so I was like you know what I am not playing this genetic lottery again.

Guilt

Feelings of guilt were also discussed. Here, guilt is defined as an internalized feeling of responsibility for a negative event, whether logical or illogical, and whether or not the cause of the event is known. One participant discussed her feelings of responsibility for what her CYSHCN went through:

As parents we wonder all the time when something happens to our child if it's because of us. ... I beat myself up all the time ... about you know, well I'm probably the one that did this to them, especially with ... my middle [child], she went through so much. ... And because ... nobody could give me a why. So, ... when our children are diagnosed and nobody can give us a reason or anything, then I don't know if it's natural, but I know I went through it. It must be my fault then.

Regret

Regret was also reported by several participants. While similar to guilt, regret is defined here as a desire that something in the past had gone differently, which results in feelings of sadness. One participant described an occurrence over which she feels regret:

So, ... I really regret that I listened to [that doctor] because clearly he was wrong, and I knew in my heart he was wrong, ... but ... I really can't understand why I didn't push him harder earlier, but finally we did get what we needed.

Desperation

Several participants described feeling desperate to find a diagnosis, effectively treat symptoms, and/or otherwise help their CYSHCN. Here, desperation is defined as the feeling of distress for a current or impending crisis, which individuals will take action to avoid. One participant talked about a time when they felt desperate:

That good [provider] for [her], we had to go out of network for that person, we just had to pay for that person because she wasn't in network and we just, the people who were in network ... either were ... too far away or we had tried them and they weren't that good.

So, at that point we were just desperate, given what was happening with her, and so we paid out of pocket for that for several years.

Uncertainty/Powerlessness

Multiple participants described feelings of powerlessness, or the inability to affect change.

One participant shared:

It was hard, because I didn't understand it, I didn't know what caused it, and we still to this day, we don't know what causes the relapses. You never know when they're going to come; she could go to bed tonight, knock on wood, and wake up tomorrow and be full blown, you just don't know.

Righteous Anger

Difficult emotions may occur not only at the time individuals became the family member of a CYSHCN, but also during the lifespan of the CYSHCN. Multiple participants expressed frustration and offense over certain incidents, and the steps they took to handle the situations afterwards. One participant talked about their responses to the difficulties of accessing certain services:

We have regional early intervention collaboratives, and at that time they were collecting letters on SSI denials and I'm like oh yeah, we weren't accepted for SSI and actually they told us, you can't make this stuff up, that our child was not disabled enough to qualify. And, I'm like, she almost died, how disabled do you have to be? So, I was like, yeah, I'll write a letter!

In addition to the emotional responses detailed above, participants also described feeling alone, overwhelmed, fearful for the child's or youth's survival, and reported that other family members experienced Posttraumatic Stress Disorder (PTSD). Participants also described being in "survival mode" for multiple years after the birth of a CYSHCN. This experience was also described as being in "fight or flight," being "in crisis," being "unable to breathe," and being a "robot."

3.3.2 Working with Providers

During interviews, participants shared experiences about different providers that they had worked with over the course of years. Multiple types of providers were discussed, including doctors, nurses, therapists, teachers, principals, members of school boards, school district administrators, employees of county departments, and other service providers, such as medical technicians and staff from the Deaf-Blind Project. The participants described both positive and negative experiences across the range of providers. Several subthemes emerged, including expectations and hopes for the CYSHCN, care coordination, and statements regarding genetics specifically. Each of the subthemes is described separately in the following section. Here, general comments on working with providers are described.

First, accounts regarding physical health providers are presented. One participant described an event in which a healthcare provider respected their input:

The surgeon came out and told my mom and I that he wasn't able to repair [what he went in to repair]. So, when we went back to talk to her with him, I pulled him aside, and I said you know what, she was really nervous today, she was anxious and if you tell her now that you weren't able to do what you went in there to do that's going to really upset her. Do you think we could wait, ... just leave it more general and I said then we can talk to her about it after she's had time to get over sort of the trauma of the surgery? And he struggled with that, and I think it's because he's been always told you tell the patient, you're always honest and direct with the patient, but then he did, he did do what we wanted.

Not all accounts were quite so positive. Several participants told stories that either they had experienced themselves or that caregivers of CYSHCN working with the family-led organizations had experienced. These encounters involved healthcare providers not listening to or believing caregivers, not supporting or communicating with caregivers, and/or not being involved enough in the care coordination of the CYSHCN. One participant described events where needed support from a healthcare provider was lacking:

She did have a pediatrician that was just her regular doctor, really nice guy, but ... he was one of those let's wait and see, so he was very much not overly concerned about anything, and that was not good for us because there were times we missed the signs that [she] was actually really ill, and we didn't know it because we were kind of his wait and see attitude, and ended up in the hospital sometimes for months on end.

Second, exchanges with the education system were mentioned by participants, which included both positive and negative experiences working with school personnel. Participants described interactions that they had experienced themselves, as well as interactions experienced by individuals who they had worked with through the family-led organizations. One participant reported:

The nurse that was at that school left in the middle of the school year and ... I guess there was no plan to get another nurse. And, so they said, well just in case something ... would happen to your daughter, we're going to need you to come to the school. And she's like well, what am I supposed to do at the school, and they're like oh, we'll find you a place you can sit in the cafeteria, or something. ... And, after she went the first couple days she was like, I have to get back to work. ... She told them you guys need to find someone, a nurse that can come in here. My daughter deserves an education, this is not her fault, and I understand that it's not your fault that she left either, but this needs to be fixed. And at first, they didn't do anything, ... the school didn't, and she went to the school board and finally they got a nurse in there.

Several participants discussed working with teachers of the CYSHCN throughout their education. One participant shared:

There were some teachers along the way who really provoked her and didn't really understand, even though they were given the information, didn't believe the disability It's hard enough for I think ... youth and young adults to understand and accept their disability and then when you have people that are telling you that it's not really a disability, ... it was devastating to her.

Third, mental health providers were mentioned. The professional skills of a mental health provider were integral in the progress of multiple CYSHCN. One participant described the improvement made when the family found a skilled mental health provider:

Once we figured out what was going on, then we were able to get her to a different therapist who understood trauma more effectively, and also adolescence ... better than the previous therapist that she had had, and worked really well with her and with us and gave us tools

that we were able to use at home to support her, so that was a very good experience. But it's unfortunate again that it took a while to find a good person, with trial and error, ... and once we started going to this other person it was like, oh my God, if only we had had her three years ago.

In certain cases, county and school district staff were identified as being supportive advocates, assisting students in finding careers that were gratifying, as well as working with teachers to enable students to complete home instruction and go on to graduate with their peers and attend college. Lastly, other service providers were also discussed during interviews. These personnel included transition coordinators from the county, staff from the Deaf-Blind Project, and others, for example, speech therapists. Again, described experiences were both positive and negative. The participants expressed that there were lasting effects from these interactions on both them, as well as on the CYSHCN.

3.3.2.1 Expectations and Hopes for CYSHCN

One underlying issue consistently mentioned throughout the interviews was that of low expectations and a lack of hope for the CYSHCN communicated to family members by healthcare providers (as well as teachers and other personnel). There were multiple accounts of both participants and the people they were helping through the family-led organizations being told that their child would never walk and would never talk. Several participants reported that not much hope was given by many physicians. For example, one participant stated:

I actually just talked to a family whose child ... does have a genetic condition. ... She said that she listened to the doctors and took him home and she realized when he was about three months old that she wasn't talking to him, ... and she thought what the heck am I doing here? And, but it was because the professionals, she listened to what they said.

Many caregivers of CYSHCN make decisions about their child's care based upon the information presented to them by physicians. Some leaders of family-led organizations discussed

the goal of connecting with families with CYSHCN when the children are young. One participant reported:

One of the efforts that we really are focusing on that I think is an important one is to get more families whose kids are young involved, because even when a child is in early intervention, getting ready to transition to school age, they may, if they just listen to professionals, they may get diverted down a path that doesn't offer them as many opportunities.

In spite of having low expectations of the CYSHCN communicated to them, several families discussed the evolution of their attitudes toward the child or youth. Other families were influential in furthering expectations the family members had for the CYSHCN. One participant described the influence of seeing other CYSHCN who were older than her CYSHCN:

... We were told lots of things, she wouldn't walk, wouldn't talk, would live in an institution, wouldn't be able to do anything. So you hear a lot of those things and you believe it, because the professionals tell you that, and so you believe that they're telling you these things, and then you begin to see people that are older than your child and you're like well, wait a minute, they're working, or they're riding a bicycle, or whatever these things are, and so you begin to broaden your perceptions of what disability and abilities are and can be.

Participants noted that some healthcare providers encouraged the family members of the CYSHCN to have higher expectations and hopes for the child or youth. Nurses were mentioned as integral to the support of the family members of the CYSHCN, in addition to the child or youth. As one participant shared:

The nursing staff were really the ones who talked a lot to us about high expectations and about not treating your baby as a kid with a disability, but treating your baby as a baby, as a person. They really instilled some things that later on we could go back and appreciate and say wow, that really made a lot of sense, because you're so ... we're like we have to know every single diagnosis, and we have to be able to spout those off every time we talk to somebody, and they're like, no you don't, just talk about [your daughter].

3.3.2.2 Care Coordination

Care coordination, a component of the medical home model that emphasizes the partnership between the physician and the family members, was discussed by multiple participants.

Both this model and the work of the family-led organizations focus on the importance of caregiver input in the CYSHCN's care. Several examples of the dismissal of family input by healthcare providers were described. One participant shared:

I took her to the emergency room, and they were like oh, it's probably allergies because it was summer time and the day before she had been at the park all day, and just that motherly instinct kicked in and was like I'm not going to keep drugging my daughter with Benadryl because I didn't feel like that's what it was.

Examples of successful care coordination were also described by several participants. This participant continued to describe her experience:

So, I ... called her pediatrician ... I was like you know ... can they get her in there's something wrong with her I don't know what it is, but there is definitely something wrong with her, just, I know it. And so, they were like bring her whenever you can, so I of course went straight there and the doctor was passing us as we were going into another room, he looked down and was like [girl's name]! He was like get [this test]... And then, it wasn't even ten minutes later, and he was coming in and saying they're waiting for you in Children's in [another city], we need to get her there, she's in risk of [organ] failure.

Another participant also talked about her experience with successful care coordination, in this case provided by a specialist. This participant said:

So, the neurologists in that practice became probably more important to us than some of the others because we needed to figure out how to help [her] be able to get through her days without having all these seizures, and so ultimately, they became her medical home. In an odd twist, we just happened to have a neurologist who has so much more wisdom and experience than any other professional we had ever been around, outside of that developmental pediatrician.

When asked about finding specialists who could really understand and connect with CYSHCN, and the general access to those services, including barriers to access, care coordination, and follow-up, one participant commented on the general state of care coordination by stating:

Care coordination doesn't exist.

Care coordination was discussed in most of the interviews. When asked about their story from the perspective of a family member of a CYSHCN, one participant concluded her narrative

with the statement that she herself performs a lot of her family member's care coordination. This participant then stated:

Care coordination just doesn't happen naturally, I think, in many medical practices.

Suggestions were also presented for healthcare providers to better work with family members of CYSHCN and CYSHCN themselves, as well as to successfully provide a medical home for them. One participant provided the following suggestions to improve the alliance between healthcare providers and family members of CYSHCN.

It's good for families to be as informed as possible, but I think it's really important for physicians to work with, meet with, interact with families of kids who do experience some of the genetic challenges, genetic issues as they're older. ... So, I think you can get a really narrow perspective if you're only interfacing with families that are thinking about having children, or families who are pregnant, or right after birth. And, see what's possible. I think the other thing that's ... a really effective practice is to partner with parents, but I don't think that that's a natural thing that happens in a lot of the medical community, and it's part of that whole medical home piece. Where we see the practices that have the medical home model in place, they're a lot more attentive to the whole person, the family, the family perspective, but I think we still have a long way to go.

3.3.2.3 Experiences with Genetics

Participants were asked about their experiences with genetic healthcare providers. They discussed personal experiences, their philosophies on genetic testing, and their experiences working with other families through their family-led organizations. The following participant discussed the diagnostic odyssey she and her family went through for their daughter, and the outcome of her genetic testing.

So, we had a [genetic test] done ... and it was found that she has ... a [genetic condition]. So, it kind of explains the constellation of things that she has going on. [It] didn't really change anything, it just kind of brought it together. ... So, when you look at some of the things that could happen with that [condition], ... it was like ah hah! ... But it gave it a name. ... I mean it was just like one thing after another for three years, and so, they said something's got to be going on. And that's when they did it, because they couldn't figure out ... we had so many different specialists, but it was like, why does she have this and this and this and this and that and nobody was connecting the dots. I mean, we had infectious disease in, we had everybody in, everybody was trying to figure out what was going on.

Multiple participants discussed the benefits of genetic testing, specifically referencing a better understanding of their options going forward. One participant shared:

We decided we're having one [child] because honestly we can't even have a dog because the poor thing wouldn't get any attention at all, because our child needs all our attention, so when you have multiple children that need that kind of attention, it's really a strain. So, I mean, that's why we decided not to have any more. And, that I think is one of the key things about genetics. The other key thing is once it has a name and you know what you're dealing with you don't feel so helpless and hopeless. It gives you some options for ok, what are next steps?

Multiple participants also stated that they felt the decisions of whether or not to meet with genetic healthcare professionals and/or to get genetic testing were personal ones. One participant described their exposure to others making decisions regarding genetic counseling and/or testing:

They were really struggling to understand each other's opinion, because one of them was saying, oh I think everyone should be told you should go for genetics, it doesn't hurt anybody to go and do this, and the other one, who has a genetic condition, said but my daughters didn't inherit it, only my son, and ... I don't want him to feel ... them to have a bad relationship as brothers and sisters because he inherited it and she didn't. So, it was so interesting sitting at this table watching them kind of argue about it, and at the end of the day they were saying the same things, that it's really a personal choice.

The lack of genetic literacy and the growing popularity of ancestry testing in the general population were also discussed as problems. One participant stated:

There's so much right now with that whole DNA ... ancestry and that people are getting confused about the two and we have had some conversations with some families to say, we're not telling you you shouldn't go out and do ancestry DNA. ... It doesn't tell you if you have any diseases or disabilities or any ... like, it doesn't say autism, Alzheimer's, it doesn't tell you anything. All it tells you is you might have been from African descent or whatever, European. ... It's unfortunate this has happened because I think people don't take the seriousness of the genetic testing and genetic counseling like I think they should, they think it is going to tell them something about where they came from.

Regarding their work with family-led organizations, several participants explained that many of the individuals they serve may have a genetic condition, but do not know if they do. The family-led organizations work with individuals with blindness, deafness and hearing loss, and autism, which often have genetic components. One participant stated:

Now we know more and more things that are genetic conditions that we didn't know were genetic conditions in the past.

Several participants also explained that although their organization serves CYSHCN with genetic conditions, the diagnosis itself is not generally the purpose behind the work with the organization. One participant discussed their experiences with the individuals reaching out to their family-led organization with issues that involved genetics:

So, they're usually calling us because there's a problem and then when we go to do the intake that's when we will find out about if it's a genetic disorder. So, we note that, and everything's fine, but the piece that we spend a lot of time with families understanding is, is when you're talking about healthcare and these are school-aged kids, it's not about the genetic disorder itself, it's about what is a manifestation because of the genetic disorder? So, if the genetic disorder is such that a kid has an intellectual disability, then what is it that we need to do to help you around the educational piece around the intellectual disability

3.3.3 Being a Leader in a Family-Led Organization

In discussing their leadership roles within their organizations, the participants covered several topics, such as their motivations, how they help others through their organizations, leadership styles, and advice they would have for new family members of CYSHCN, including those who want to get involved with these family-led organizations. Each participant had their own story of how they became involved in their family-led organization, which began with the personal experience of being a family member of a CYSHCN. The amount of time between becoming a family member of a CYSHCN and becoming part of a family-led organization varied substantially. Several individuals first went to a family-led organization for the support and resources it offered, others began their involvement by participating in advocacy work, and still others by becoming employed by a family-led organization.

Multiple participants described the process of learning, advocating, meeting other advocates, and then being invited to participate in different advocacy organizations, including family-led organizations. References to benefits of both having mentors and being mentors to others were repeated across the interviews. The participants also discussed how their involvement had evolved over time. One participant stated:

I was like, you know what, I really need to round myself out a little bit more so I went to work at a charter school doing special education, teacher development, professional development, and stuff like that, but what happened was, it was a kindergarten school, “K” through five, and I ended up working with kids and it just tore me up. I was like, yep I really cannot be in the trenches ... So I left that position and at that time I ... went back and got my masters ... and, because I realize I also, not only did I not need to be doing front-line work, but I also really said ... we really got to look at some systems change, ... and we have to start young.

Several leaders said that the motivation for their work was their desire to help others in the same situations as their own family. One participant discussed the importance of helping others through their family-led organization:

And then I found this, and I love this. It’s important to me personally, it means a lot to help the families because I didn’t have a lot of that, I did a lot navigation and a lot of figuring things out on my own, ... it took me learning to do it myself. So, I try my hardest with the families that I do get to help them with anything and everything that’s in my power because I didn’t have that, so I love giving that to other people, and letting them know that it’s out there, and you can have the help.

Each of the leaders described different ways in which their organization worked to help families of CYSHCN. These approaches included training both the family members of CYSHCN and the healthcare professionals. One participant talked about strategies in working with family members of CYSHCN.

For the most part we deal with the parents and trying to teach them to advocate for themselves and for their child’s needs, teach them that this is a partnership and that you need to know what’s going on with your child and try to definitely get the doctors and the school staff to understand, I know what’s going on, this is my child, ... that my voice matters.

The same participant also talked about strategies in training healthcare professionals to better work with CYSHCN and their family members:

We take our children to doctor and they say ok, these are the medications or [this] is the equipment that you need to use and learn and this is what's wrong with your kid, all of this stuff, but they don't know about the home life and how difficult sometimes home life can be. So, ... that's what we want all doctors and staff to see is that they are more than just their diagnoses. And there's other things going on, and to also look for those signs, like if the parents come in and they're not themselves, to ask like, how are things going at home? Is everything ok? Because, people that have children with special health care needs have high percentages of depression, and suicide, divorce, anxiety, all of these other things. So, we need to see it as a whole ... as a family.

Different leadership styles were also discussed; these incorporated diverse strategies for both training the individuals who work for, and serving the families that come to, these family-led organizations. One participant first shared her viewpoints on strategies for training individuals who work for the family-led organization:

So, I really think we as parent organizations have to spend a lot more time mentoring young parents to be ready for this stuff and not pushing them in too quickly ... and so I really think we have to take some time and make sure that parents are ready, and it's not our job to say when they're ready, it's us to say we think you will feel ready, you will feel no longer in fight or flight, you will feel like you can truly listen and absorb and be unbiased.

The same participant also shared her viewpoints on strategies for serving the families that come to the family-led organization:

I wanted everyone to kind of have that same philosophy of around how we're supporting parents in getting information. That it's not our place to tell them what to do or how to feel, it's our place to use a lot of humility and empathy, and give them hope, never tell them it's going to be ok, because we don't know what could happen, but at least if we give them hope that there are possibilities.

Several individuals also discussed challenges they faced as leaders of these evolving organizations. Some of these challenges included reaching underserved and/or vulnerable populations, connecting with new families in light of changing preferences for communication,

and dealing with fragmentation in both resources and systems. Participants also discussed fundamental challenges within the sphere of family-led organizations. One participant stated:

It's quite depressing. I've been doing this for almost twenty-nine years, and as I hear from my team members and my staff about they'll come in and they'll ask me, ... what do I do about blah, blah, blah, or we just got a call about blah, blah, blah, and I'll look at them and I'll go, these were the exact same issues almost thirty years ago that we were still dealing with. It's just so disheartening that it hasn't gotten any better. Yeah, it's so disheartening. ... We sit as directors and have these conversations, and we're so disheartened because it's like it hasn't changed any, it's like it hasn't gotten better.

One fundamental challenge is the potential conflict between emphasizing the importance of a diagnosis and at the same time emphasizing the importance of acknowledging the CYSHCN as an individual first. One participant described facing this challenge as a leader in a family-led organization:

I know in the Parent-to-Parent world you want to try to find an exact match, you want to be able to say oh my goodness, [she] has the exact same disability, and as a parent you value that because you can say they walked in my shoes exactly, they know what this is, but there's another part of it that ... at the end of the day [my daughter] is [my daughter], and so there's a part of me that wants to say to parents I know you're concerned about these things, and there's a piece of this that you've got to figure out what's going on, you've got to know what are the long term repercussions of this particular thing that might cause learning disabilities, or might cause seizures, or whatever it's going to do.

This participant went on to describe the other aspect of this issue that causes dissonance in their underlying principles:

But also, they're a kid, and can you get to know who they are and their gifts and talents and they're part of your life, and so if you get so focused on trying to get a diagnosis and understand every component of [her] seven or eight disabilities, you get to a point where that doesn't matter anymore, what matters is you got to parent this kid just like you do your other kids, you've got to make sure that they know how to behave the way you want them to behave, when they're out in public, like you would do with any of your other kids, you want them to learn the life lessons and dignity of risk and if they do this then this might happen, you can't wrap them in bubble wrap, and so there's a part of me that struggles sometimes ...

Lastly, participants discussed advice they would have for individuals new to being family members of CYSHCN, and for family members of CYSHCN and CYSHCN themselves who

wanted to get involved with these family-led organizations. One participant discussed their advice for family members of CYSHCN:

I think the first thing to do would be, is one: take care of yourself first, so what do you need to learn for yourself, what do you need to learn for your child, and you need to take care of yourself and become your child's best advocate first and foremost. And that's one of the ways you can become involved in the organization is by learning, because then you know our work and you can speak first-hand to our work. Then, I think after that, depending on if folks feel like they can handle that piece, is volunteering, there's lots of volunteer opportunities that families can do and it can look a lot of different ways, including serving on a board.

Another participant also shared their advice for family members of CYSHCN interested in getting involved with family-led organization work:

I think having the opportunities to take leadership opportunities at whatever level you can, so to start off even small, but don't underestimate the power of that family perspective. I think that learning to tell your story in such a way that it is powerful, and it is a really helpful way to influence people, so I think learning to tell your story, and I think also the listening skills and collaboration is really important because there's many, many organizations, many nonprofits, and ... you're likely to be more powerful if you can find allies and work together than trying to take on everything yourself. I think just not to underestimate the power of the lived experience that someone has and trust your instincts.

3.3.3.1 Issues of Access

Participants were asked about the barriers to access to care, such as physical healthcare, including genetic services, mental healthcare, and other services, such as speech therapy. Statements were also made about access to information, health insurance coverage, and equipment and prescriptions. Difficulty with financing the care of a CYSHCN, as well as successfully connecting individuals in underserved communities with the family-led organizations were also discussed. One participant talked about specific work that was done to address issues related to communications access:

Some people don't have very good internet access, and their phone is their only internet access, and it's hard to print and read when you just have a phone, so a lot of the things that I do for people are, print that kind of research, like medications, and side effects, and

things like that. I've researched different places people can take their kids for behavioral support, or stuff like that, and I don't recommend anybody I just send them a list of people in their area where they can do what they wish.

Both the difficulties of underserved communities being able to access care, as well as the challenges encountered when trying to successfully reach underserved communities were discussed. One participant discussed multiple challenges and strategies in reaching underserved families:

It's a challenge to serve all the families in the state, to reach underserved families, because we have underserved families in rural areas and then we have a different sort of underserved family in the urban areas. ... We're looking at wanting to use data to take a look at are we reaching those underserved areas? How can we be strategic in both responding to requests, but also outreaching to partners like, we've not been in this county to do any training in two years, let's make sure we outreach and try to do that. ... I think one of the things that's really challenging is the ruralness of [the state]. So, there aren't necessarily advanced medical care facilities or physicians that have specialties located in some of the more rural areas and so, the very first challenge is finding a doctor that you can get to, and the trials of having to drive all day, and stay overnight sometimes even.

There are additional underlying factors complicating the care of individuals in certain underserved and/or culturally diverse populations. One participant outlined some of these factors:

One of the things about working with underserved families, whether they're immigrant or African American or low income, is that, well two things, one is that their children often aren't identified as early as White children are, or middle-class children. So, one reason is that if you go to a federally qualified health center, or a community clinic for your child when they're young, ... you're not seeing the same doctor every time, like you are if you have, here's my primary care provider, I've got regular insurance, ... and [the doctor] can see that my child isn't developing. ... The other thing is the cultural implications of disabilities and special health care needs in diverse communities. ... There are very, very many cultural barriers to families either accessing evaluation to identify if their children have special health care needs or accessing services if their children are identified. ... And so, having staff who are from that community who can tell their own story about why it was so important ... to get their child screened and to follow-up the screening, get their child evaluated and then to access services is a very powerful and effective tool.

One vulnerable subset of the underserved population is that of immigrant families. In addition to helping families navigate physical and mental health systems, the education system,

and connecting families with support networks, some family-led organizations also address the broader concerns of members of immigrant communities. One participant reported:

Well, a lot of the good work we did in immigrant communities was undermined by the current administration. The year before the current administration came into power, we had this six-session ... resource parent training that we do every year, ... the year before that, the Spanish training we had had forty-five parents at it. The year after that election we had five parents. ... We've also had situations where we've had school staff call ICE [Immigration and Customs Enforcement] on parents, which you're not supposed to do. ... Really this is something that's ... affecting not just undocumented immigrants, but also immigrants who are here with papers, and even people that have a last name that is Latino. So, we had a parent call us, a parent who's a U.S. citizen, whose child was born in the United State, just has a Spanish last name, call us because their five-year-old was told by another five-year-old at the playground, we're going to build a wall and throw you on the other side of it. So, it's not surprising that that population is very concerned and has a lot more fear that we had helped them overcome

Large expenses of caring for CYSHCN and loss of income were also discussed as both barriers to accessing services and a significant drain on financial resources. One participant talked about large expenses and their downstream effects:

And, we were saying, we think we're going to have to file bankruptcy, we are drowning in debt from the hospital, not all the bills were paid, we're drowning from ... she was starting to have early intervention services, and early intervention in our state at that time you had to pay out of pocket, ... and she had 6 visits a week, and so ... everything was just like, six hundred dollars a week for one hundred dollars a visit ...

This participant went on to talk about the challenges of care, including loss of income, as their family member's ability to work was affected:

It did require that my husband not work, so that was another challenge, I think for us as a family, is one of us has to be at home with her, and I used to work out of the home, and we had some child care and some school care for [her], so I could go out of the house while she was out, ... and so, that's another unfortunate reality of when kids get a certain age, there's no childcare for them after the age of twelve years old or something, and so ... one of us has had to be with her every day, and so it means one income, because he has to be here with her.

Participants discussed the requirement that they be flexible in accessing systems and services, and how this also affected their ability to work. They specifically described not being

able to work, or work full-time, and the challenges of taking care of a CYSHCN and being employed at the same time. One participant shared:

I ... was a stay at home mom once she was diagnosed. I stayed at home because it was really hard getting a job to work with me on doctors' appointments, and taking her for lab work, and sometimes the school couldn't get her to take her medications so I would have to go to the school to make sure she took her medication and she had to be on a special diet and sometimes that involved me taking lunch to her and things like that, so it was just easier for me at that time to be a stay at home mom, but then when the relationship broke up, ... I had to turn into a working mother.

System fragmentation was also listed as a barrier to receiving care, as underlying infrastructure between complex systems was absent. This fragmentation has been illustrated by both poor communication and the lack of joint effort within and between systems, including healthcare and education. Multiple participants described system fragmentation as affecting financial resources, available care, and effective communication across all services. These effects can be seen in healthcare and education systems, support services, and the connections between them. One participant discussed the impact that system fragmentation has on how they can help families of CYSHCN through their family-led organization:

The systems ... that don't work together, so we can pay with a waiver for nursing but there aren't enough nurses that do home care nursing to support kids that have those complex health issues, and so there's barriers that way, ... or we can pay for nursing but ... there's no funding for the ramp you need to be able to get the wheelchair in and out of the house, that has to come from a different funding source, and those funding sources don't work together.

Specifically, fragmentation turns what could otherwise be routine and coordinated care of a CYSHCN into a multi-step process that needs to be managed by the family members of the CYSHCN. One participant shared:

[The doctor] would look at her, and they'd be like, well we have to get a hold of [her specialist] before we can prescribe anything. So, a doctor visit that could have been like, ok, here's an antibiotic go on about your way ... turned into a way bigger situation because I'd have to take her home and wait for those phone calls, wait, and possibly then, some ... a lot of times [her specialist] was like, oh well, we need to see her here. So, on one day

I'm taking her to her pediatrician because she has the sniffles and a cough, and then I'm driving three hours to [her specialist].

Multiple participants detailed the challenges of working with health insurance companies to obtain equipment, access services, and receive care coordination. One participant stated:

She and her husband had to really fight the insurance company because when he was four, he really needed a power wheelchair to help his independence. And ... we don't provide those for four-year-olds, is basically what they said. So, they had to do all kinds of advocacy, and ... you should see this kid zoom around in his power wheelchair.

3.3.3.2 Progression of the Family-Led Organizations

Each participant was asked about the family-led organization they work for and how it has evolved over time. One subtheme emerged during the course of the interviews, that of ideological differences among organizations and their impact on the alliance between family-led organizations; this subtheme is described below its own section.

In addition to endeavors previously mentioned, participants talked about planning and development work they had done to fill gaps when there was a need for services. One participant commented:

We did a strategic plan with the board, to try to look at again, ... where should our emphasis be, ... there are so, so much need, how do we most effectively meet the need?

This same participant commented on completing some of the organization's work in collaboration with others:

We do a lot with partnering with other organizations, cultural brokers to help us get connected with underserved communities.

Several participants discussed work their family-led organizations had done to meet these service needs. One participant stated:

We just worked with our state to develop a shared plan of care that was piloted with federally qualified health centers, who as you know, serve the most underserved families and the families that are least likely to actually have access to good care, and to specialists,

and to good care coordination. So, we've done a lot of kind of collaborative work with professionals.

The participants were asked what questions or reasons individuals usually have when contacting the organization, and anything else about the organization in general that they wanted to share. The leaders in the family-led organizations discussed specific work that was done directly with families and also how the organization works to serve them. One participant stated:

Typically, ... at the end of the day it's families aren't feeling like they're heard and either they're trying to get an evaluation and they don't feel like ... they're not making any progress with it, or they have an IEP that's not being followed, and there are health concerns and health issues that need to be included in the IEP and those aren't, so that kids can access their education. So, it's all kind of mushed together.

The work that the organizations do to adapt to the changing needs of families with CYSHCN was also discussed. Two of the challenges of connecting with families of CYSHCN are that they are very busy with taking care of their CYSHCN, and also that many individuals currently contacting family-led organizations prefer to communicate through text or email. One participant discussed using technology to adapt to the challenge of connecting families of CYSHCN to family-led organizations:

It's hard for families to commit to training, day-long, like we do lots of leadership trainings, because of demands and things and competing priorities, which I get that, but when they get there the connections they make with other families is so powerful that I don't ever want to lose that, ... and then once they make those connections, then they go to using technology ... to stay connected. ... So, we have to be responsive and I can't please everybody, but we are trying something that's going to be more responsive.

This goal of making meaningful connections was commented on by several participants. One participant discussed the associated challenges of text message and email methods of communication:

We're seeing probably more so that people want us to text them and just give them a quick answer, and there's that disconnection when you don't have when you're on the phone you can still hear the inflection of a voice, when you're face-to-face, when you're on video, but if you're trying to do this through text and email there's a wall there, you can't really

build that connection, and again I'm a relationship person, I like to be able to build a relationship and it's hard to do that through five text messages that are going back and forth really quickly.

This same participant also discussed differing preferences across cultures for certain methods of communication:

There are still parents who want face-to-face, they want parent support groups. We work with the Spanish community, and it's very culturally appropriate to do stuff in groups and so they still come together. The Arabic community ... we have some staff who are Arabic and ... they still tend to come together as a group and talk things out over food and have conversation.

3.3.3.3 Ideological Differences Among Family-Led Organizations

When asked to talk more about their involvement in the organization and how their participation had evolved over time, some participants expressed their opinions about fundamental aspects of family-led organizations, including the importance of collaboration. One participant shared:

I am a firm believer that it's all about relationships, I don't care who you are, what you do, what business you're in, if you are honest and humble and you are collaborative and you agree to disagree but you do it in a polite way and you know everyone has their viewpoint and you can brave into those hard to have conversations in a ... and have conflict in a way that's not ugly, people will want to work with you, people will want to reach out to you, and so I'd like to think that's one of the reasons why we have the success that we do in the work we do is because I built so many relationships.

In addition to discussing collaboration, some participants also talked about the missions of the family-led organizations in relation to working to obtain funding. One participant shared:

... Those are two new grants I was able to apply for and get for the organization since I've been here ... so that really was a nice boost. And they fit with our mission, so we're really careful not to ... we don't just go after money unless it fits in our mission and values, what we're trying to accomplish. The other thing I think I've done more of is really worked on establishing collaborative relationships with other organizations and governmental entities. Sometimes, I think when you first start something, and you're fighting a lot of battles to even be heard, ... you can end up in some adversarial relationships, and so [the organization] always had a really good reputation for knowledge and stuff, but sometimes, the collaborators were a little, maybe, mistrusting. And I was lucky enough to have worked as part of, sort of, the system for the thirty-two years I did, and so, I built a lot of

relationships there, and I was able to build on those, I think. And I think I've been able to expand ... the positive perspective that people have of [the organization].

Lastly, conflicting viewpoints regarding the fundamental aspects of family-led organizations were expressed among different participants. One participant stated:

There are groups who promote their family organizations and who talk some trash about professional organizations, and how you know, we know better we're parents, and I'm like, yeah it takes everybody, I don't know everything, you got to have the professional with the scientific knowledge, you got to have me with some wisdom, and you got to have my kid, who has her own histories and narratives, and you put the three of us together we're awesome, but you can't do it without all three of us at the table.

Another participant stated:

... Family-to-Family Health Information Centers don't have to be housed at a family-led organization. ... [The] other kinds of grants that parent centers apply for, and the Family-to-Family where Family-to-Family law doesn't say you have to have a board the majority of which is parents, that's where we're seeing these ... other organizations that really haven't been in the parent engagement field applying for competitive grants that family organizations had traditionally gotten, and now getting them themselves ... in part because they're professionals. They pay for a grant writer, and they maybe have an independent evaluator written in, because they have a lot of staff, there are different reasons why this might happen, ... just because they were rated more highly definitely doesn't mean they're going to be able to do as good a job as a family run organization can because it's not their mission. [The other organization]'s mission is not empowering families to advocate for their children and to become leaders in the system. A family run organization, that's our mission, and so, ... that's a troubling development that we've seen.

3.4 Discussion

Leaders of family-led organizations have a wealth of knowledge applicable to integrating family-led organizations into systems of care and incorporating the medical home model, as well as a unique perspective. The positive effects of the unique type of support offered by family-led organizations have been documented largely from the perspective of the parents of CYSHCN. By interviewing leaders of family led organizations, the scope of this currently limited area of research

is increased. These leaders have a unique perspective of being both a family member of a CYSHCN as well as a leader in a family-led organization. These interviews explored their experiences and evolution, using a method where they represent themselves directly. This study investigated the barriers experienced and addressed by leaders of family-led organizations.

Thematic analysis of the interviews of leaders within family-led organizations identified several themes and subthemes. Themes included Experiences of Being a Family Member of a CYSHCN, Working with Providers, and Being a Leader in a Family-Led Organization. Subthemes included Difficult Emotions Associated with the Experience, Expectations and Hopes for CYSHCN, Care Coordination, Experiences with Genetics, Issues of Access, Progression of the Family-Led Organizations, and Ideological Differences Among Family-Led Organizations. A detailed list of these themes and subthemes is shown in Table 5.

These themes and subthemes will be discussed in the following groupings: first, the experiences of being a family member of a CYSHCN, and difficult emotions associated with the experience; second, working with providers, expectations and hopes for CYSHCN, care coordination, and experiences with genetics; and third, being a leader in a family-led organization, issues of access, progression of the family-led organizations, and ideological differences among family-led organizations.

3.4.1 Theme of Experiences of Being a Family Member of a CYSHCN and its Subtheme

In both the descriptions of services offered by family-led organizations to family members of CYSHCN and the research studies of parents of CYSHCN, the unique experience of being a caregiver to a CYSHCN and the resulting psychological effects are communicated.^{3,65} In its description of family-led organizations, the National Genetics Education and Family Support

Center lists multiple sources of support for these families in dealing with not only healthcare and education systems, but also with emotional and mental health issues.³ They explain that connecting family members with services is sometimes not enough, and specific supports are needed.³ The family-led organizations can connect family members of CYSHCN with condition-specific support groups, one-on-one matched support, and support for the mental health of the individual family member and/or family as a whole.³ This may be especially important as one study by Pelentsov, et al. from Australia reported that the results of an online survey taken by parents of children with a rare disease indicated that 46% of the parents surveyed felt desperately lonely.⁶⁵ Additionally, 37% of parents were being treated for depression, 41% for anxiety, and 10% for other mental health issues.⁶⁵

The complex psychosocial progressions reported by the participants following the instant they became family members of CYSHCN are consistent with previous literature. They may have felt loss for the unaffected child or youth they thought they would have, and possibly may have also felt loss for additional unaffected children in the future. Participants reported feelings of desperation, regret, uncertainty, powerlessness, and anger, as well as feeling alone and overwhelmed. While these emotions do not seamlessly map onto the stages of grief, they are consistent with bargaining (in feelings of desperation and regret), anger, and depression (in feeling alone and overwhelmed).

In discussing grief reactions, the stage of bargaining is sometimes substituted with the stage of guilt. As stated by Kessler, et al., “guilt is generally associated with responses of self-reproach to violations of internalized standards”^{74(p675)} and may be followed with negative self-evaluations.⁷⁴ Kessler, et al. also reported that personal responsibility (or guilt) can be a defense against powerlessness in that it infers some level of control.⁷⁴ There is likely an interplay here

between the psychological reactions of bargaining, desperation, regret, and guilt, and powerlessness, although future research would be necessary to confirm this theory.

As true denial is rare, a mimic of denial may instead be an initial grief reaction; two of these mimics are disbelief and deferral.⁷² Disbelief is characterized by confusion and deferral is characterized by avoidance of implications; the basis for these differing responses is their origin: disbelief has its origins in dissonance regarding information given and deferral has its origins in the lack of physical or psychological resources.⁷² Several participants expressed their initial feelings of being a robot, unable to breathe, in shock, in fight or flight, and/or in survival mode. It is possible that at this time the participants were experiencing disbelief or deferral, although more data would be needed to support this speculation. Lastly, there is (possibly) acceptance of the new normal, which is discussed below.

An individual will undergo both defense mechanisms and coping strategies as they work through the psychological consequences of a loss.⁶⁹ Defense mechanisms are unconscious methods and coping strategies conscious methods for the mind to navigate the emotions after a negative event.⁶⁹

While there is no official consensus on a definitive list of defense mechanisms, they do include intellectualization, which is an intellectual (instead of emotional) response, and sublimation, which is the shifting of an emotional response from a socially unacceptable outlet to a socially acceptable one.⁶⁹ An example is when “pain and anger over a child with a serious disability are channeled into committed, thoughtful work for a parent support group.”^{69(p9)} The defense mechanism of sublimation may bring family members of CYSHCN to work with parent support groups. Participants’ motivations for working in family-led organizations are discussed below.

While different coping strategies are not mutually exclusive from one another, there are several identifiable strategies, which include both information focused and emotion focused, where emotion focused coping strategies tend to occur in situations that are unchangeable.⁶⁹ Lastly, there is the coping strategy of reappraisal, where one's perspective changes to be wider and more positive.⁶⁹ Family-led organizations may facilitate the adjustment to the new normal in several ways, including promoting the coping mechanism of reappraisal, and providing resources, while incorporating emotional support. Several participants discussed how being connected to another individual through a support group or one-on-one with another family member of a CYSHCN informally was beneficial for the perception of their situation. Family members of CYSHCN often feel more comfortable discussing their situation without fear of judgment with another family member of a CYSHCN.

3.4.2 Theme of Working with Providers and its Subthemes

Participants described both positive and negative experiences working with providers, such as physicians. One of the reasons identified for the negative experiences was that the way providers talked to family members of CYSHCN gave them low expectations and little hope. This issue is generally not directly discussed in the literature. In one article a mother describes the situation from her perception of the providers' perspective.⁶⁴ Gore Olsen stated, "Medical professionals seem to be caught in the dilemma of painting a pessimistic picture of the reality of the disease and encouraging the parents to maintain a positive attitude despite the realities of chronic illness."^{64(p517)} While the need to pass on information was acknowledged, all interactions were discussed from the point of view of the participant and emphasized the importance of recognizing the family voice. A suggestion offered by one of the leaders in the family-led

organizations during their interview was to have medical professionals meet children and young adults with some of these genetic conditions so that they would have a better understanding of what the future may look like. Another suggestion was to embrace family-centered care, so that the family members of CYSHCN and CYSHCN themselves have more of voice in their healthcare.

In one study by Beene-Harris, et al. examining the barriers to accessing genetic services, focus group participants identified provider lack of knowledge and awareness of genetic services, and coordination of care/referral as institutional barriers to care.⁵ Aspects of care coordination that were discussed included access to information, follow-up care, referral to support groups, assistance transitioning from one stage to another, and quality education.⁵ All participants in the current study discussed care coordination in some form, including whether or not it is currently provided, its benefits, and the central role the participants play in the care coordination of their CYSHCN. Care coordination is an integral part of the medical home model, which is promoted by the American Academy of Pediatrics⁸ and incorporates family-centered care. This model improves not only care of the CYSHCN, but also their family members.^{33,35} By implementing the medical home model, the burden of care coordination would be removed from the family members of CYSHCN.

Family-centered care also has the potential to improve experiences with genetic services. Promoting a partnership between providers and families encourages discussion of the pros and cons of genetic testing and ensures the family's right to decide. Additionally, care coordination may also reduce the prevalence, or duration, of the diagnostic odyssey, as the healthcare of the CYSHCN is centrally managed.

3.4.3 Theme of Being a Leader in a Family-Led Organization and its Subthemes

The leaders within these family-led organizations have experience as advocates, caregivers, learners, campaigners, mentors, and organizers. In these roles they have gone through the psychosocial processes, medical situations, and personal growth detailed above. They have both been supported by and supported others through their family-led organizations, the benefits of which are documented.^{1,33} When family members of CYSHCN are connected with parent mentors and family-led organizations, they benefit not only in improved navigation of community, education, and healthcare services, but also in improved mental health.³³ One study by Ainbinder, et al. found that supporting parents were seen as dependable and both normalized the learning parent's experience and gave them hope for the future of their child.¹ The supporting parents were not only available to the learning parents, but also the relationship between the parents was equitable.¹ Many of the learning parents expressed satisfaction at the opportunity to provide support to their supporting parents.¹ Many of the participants in the current study discussed first interacting with family-led organizations as the learning parent. Multiple participants also detailed the desire to help others in similar situations as themselves as their motivation for their work in the family-led organizations. These leaders of family-led organizations are strong resources for making positive change in improving the lives of families of CYSHCN and CYSHCN themselves, incorporating the medical home model, and improving access to services across systems and populations.

Participants were specifically asked about their interpretation of barriers to access, and answers arose that were consistent across participants. While this question was answered mainly from the perspective of being a leader in a family-led organization, participants did include responses in the context of their own experiences. They discussed ruralness, cost, limits on being

able to work, health insurance, inflexibility and fragmentation of systems, and difficulty of navigating healthcare and education systems as challenges. While unaffected children can be treated in primary care settings, this and a previous study indicated that caregivers of CYSHCN are often required to seek, and coordinate, specialty care.³³ One participant specifically described difficulty in obtaining what would otherwise be routine healthcare when her daughter had to see a specialist for sniffles and a cough. This is consistent with a previous article by Allshouse, et al. describing the issues of increased cost, missed work and school days, and the need for additional transportation and childcare (for other family members) as other burdens to the families of CYSHCN.³³ Difficulty in coordinating work and caregiving, as well as the associated financial burden, were also both mentioned by multiple participants referencing their personal stories as well as stories of those they have helped through their family-led organizations.

The progression of the family-led organizations was discussed by all participants. The organizations have evolved over time to attempt to reach more families, especially underserved families, and to fill identified gaps in services. They currently face the challenge of connecting with families remotely and are investigating techniques and strategies to do this effectively. Multiple participants mentioned the importance of seeing a person or hearing their voice, as opposed to texting, messaging, or emailing, when trying to make an initial connection.

Additionally, the mission of the organizations was mentioned several times in relation to the funding the organizations apply for and receive. While collaboration was emphasized, ideological differences about who should receive funding to perform the roles filled by family-led organizations were striking. One article by Anderson, et al. described this tension regarding grants awarded to measure quality in efforts to increase cost effectiveness of healthcare.³⁵ The authors stated, “although Family Voices and other family-led organizations received small stipends and

contracts through some of these initiatives, the majority of the funds have gone to universities or state agencies. ... There is substantive knowledge and experience among families of CYSHCN to contribute to quality improvement efforts.”^{35(pS102)} While all participants were externally identified as being part of a family-led organization, there were conflicting views on whether a family-led organization could, or should, be housed in a university. As the input of families is integral to the adoption of the medical home model and increasing access to services, disagreements on the definition of family-led organizations and who should be eligible to receive funding could potentially be a drain on valuable energy, time, and resources. Collaboration among all parties involved may be a more efficient approach to meeting shared goals.

3.4.4 Limitations

Although this study included interviews of individuals from multiple states at multiple levels in the family-led organizations, the main limitation of this study is that only six interviews were conducted. These individuals represent five of the eight states (including the District of Columbia) within the NYMAC region, and both Parent-to-Parent and Family-to-Family groups. But, because of the small total number of interviews the analysis may be missing other perspectives on the issues. There may also be selection bias between the individuals who volunteered to be interviewed, who may be more comfortable telling their stories and expressing their opinions, versus those who did not volunteer to be interviewed. Additionally, this study focused on the NYMAC service region, and so the findings may not be generalizable to other regions of the country covered by other Regional Genetic Networks. Lastly, the transcripts were coded by only one person, which means that certain interpretations may be altered if a second person were to code the transcripts. While one person coding transcripts is satisfactory for the current publication,

an additional person to code the transcripts, and then complete consensus coding, where assignment of codes is reviewed until a consensus is reached, would be necessary for further publications.

3.4.5 Future Directions

Future studies may aim to look at aspects of the caregivers of CYSHCN and aspects of the family-led organizations brought up in this study but not further explored and those not generally present in the literature. This could include an analysis of the effects of the expectations and hopes for CYSHCN on both their caregivers and the CYSHCN themselves. A detailed analysis of the emotional response of individuals (who would later become leaders) to finding out their family member has a special health care need is also possible. Additionally, further research into the nature of collaboration and partnership between different family-led organizations may help improve the efficacy of these organizations even further.

These leaders are valuable resources for improving the lives of families of CYSHCN and CYSHCN themselves, incorporating the medical home model and enhancing access to services across systems and populations. Further studies regarding accomplishing these goals would benefit from including leaders of family-led organizations in the research process. Working with leaders of family-led organizations may also benefit researching how to achieve successful connection with learning families remotely using technology. An additional area of interest may be to explore the effects of engaging in the suggestions made for the healthcare providers, including having them interact with older children and young adults with special health care needs. This would help determine if this strategy is successful at helping providers go on to speak to family members of CYSHCN truthfully, but also without eradicating their hope for the future.

3.5 Conclusions

There is much to be learned from leaders of family-led organizations, which have not been studied in full and should be further investigated. This study was small in nature, but provides a starting point for larger, more in-depth studies. Studies may include the importance of these organizations and strategies to integrate them into the systems of care. The medical home model has considerable potential to improve healthcare and family-led organizations could play an integral role in this transition. Healthcare providers, and others, such as teachers, have the opportunity to become families' champions. Lastly, attempts to improve access to services are currently underway but would benefit from additional support and research.

4.0 Research Significance to Genetic Counseling and Public Health

Family-led organizations including P2Ps, F2Fs, and SAOs are integral to the comprehensive care of CYSHCN and their family members. While these organizations serve families other than those whose members are affected with genetic conditions, if a family member has a genetic condition, these are the non-diagnosis specific support organizations they could go to for support. Both the Individuals with Disabilities Education Act (IDEA)³⁵ and the updated formal definition of CYSHCN⁹ published in *Pediatrics* focus on the individual with special health care needs instead of their diagnoses. As healthcare professionals, genetic counselors are integral components of family-centered, coordinated care of CYSHCN and their family members. It is important that genetic counselors are aware of these organizations, as the individuals they see in clinic may be in need of referral to one or more of them.

One group working to bring together leaders of family-led organizations, healthcare professionals, and service delivery specialists is the Regional Genetics Networks (RGNs) and their National Coordinating Center (NCC). These organizations are charged with linking advances in genetics with improvements in public health.²⁵ The National Genetics Education and Family Support Center is a three-year initiative that works alongside the RGNs to connect individuals and families to direct support. This Family Support Center helps the RGNs to partner with family-led organizations and reach underserved families; provide information on education and healthcare for genetic conditions; connect families with emotional support and other services; and provide training opportunities at the individual and systems level.³ Genetic Alliance is the national organization that is leading the Family Support Center initiative, as well as working on advocacy-led research, family planning, and stakeholder engagement programs. While Genetic Alliance was

originally focused on supporting individuals with genetic conditions, the organization has expanded to supporting individuals and families with diverse healthcare needs and endeavors to keep them active participants in healthcare.²⁷

There is a national infrastructure of multiple collaborating organizations whose goals are to connect with CYSHCN affected by genetic conditions and their family members, and to help them access the services and supports they need. Genetic counselors may not only benefit from being connected with these organizations in order to help the individuals in their clinics, but may also contribute to the local, regional, or national efforts to improve access and care for these individuals.

Family-led organizations are integral to all three of the core functions of public health.⁷⁹ They are often the first to know about health problems that may affect a community, fulfilling the assessment function; and they are also connected with and have insight into multiple healthcare and support services, fulfilling the assurance function. These organizations are advocates for policies that will improve the health of children, including CYSHCN, at the local, state, and national levels, fulfilling the policy development function. In addition to the three core functions of public health, there are ten essential public health services.⁷⁹ Family-led organizations provide many of these services, including connecting individuals to health services and helping them receive needed care.⁷⁹ These organizations are also vital in examining access to and the status of health services.⁷⁹ Lastly, family-led organizations satisfy all three of the policy development essential services: they “inform, educate, and empower people about health issues; mobilize community partnerships and action to identify and solve health problems; [and] develop policies and plans that support individual and community health efforts.”⁷⁹ By interviewing leaders of family-led organizations, insight may be gained into each of these essential public health services,

specifically informing the goal of increasing access of healthcare services, including genetic services, to CYSHCN, their families, and the general population.

Genetic counselors and public health officials would benefit by knowing about and being more connected to the multiple organizations mentioned here, including family-led organizations. Engaging in a coordinated effort to provide the previously mentioned essential public health services may contribute to increased access to healthcare services overall, including genetic services. By being connected with these organizations and each other, both genetic counselors and public health officials may not only benefit in their personal work but may also contribute to access and care for both CYSHCN and the general population. Areas that would benefit from a collaboration between genetic counselors, public health officials, and family-led organizations include the integration of family-led organizations into healthcare and education systems, as well as the implementation of the medical home model. Having paid staff from family-led organizations working at the same sites as genetic counselors and public health officials is one way to foster this collaboration. The greatest benefit to CYSHCN and their family members is more attainable when all these providers are working towards a common goal.

5.0 Public Health Essay: Analyses of Relationships between Demographics, Medically Underserved Designations, & Number of Genetic Clinic Sites in the NYMAC Region

5.1 Background

The health of individuals and populations is dependent upon the resources to which they have access.⁸⁰ There are many pathways by which education and income effect the social advantage of individuals and populations.⁸⁰ Information on several demographic variables related to education and income is publicly available,⁸¹ as is information on government-identified (underserved) shortage areas,⁸² or populations and places with limited access to healthcare. By also utilizing a detailed map of genetic clinic sites, the relationships between demographics, regions designated as medically underserved, and the location of clinic sites was investigated.

5.1.1 Social Determinants of Health

The social determinants of health, otherwise referred to as upstream factors, encompass the availability of opportunities and resources that impact health disparities on an individual and systemic level.⁸⁰ Both social and economic upstream factors, specifically educational attainment and income, greatly influence health outcomes.⁸⁰ This is shown in Figure 4, where the top of the figure represents the most upstream determinants, which then affect the determinants below them in the figure.⁸⁰ For example, economic and social opportunities and resources affect access to healthy living and working conditions.⁸⁰ Medical care and personal behavior, such as cigarette use, are downstream determinants that influence health. These downstream determinants are often

targeted by intervention efforts, as the relationship between upstream and downstream factors is complex and occurs over a long period of time.⁸⁰

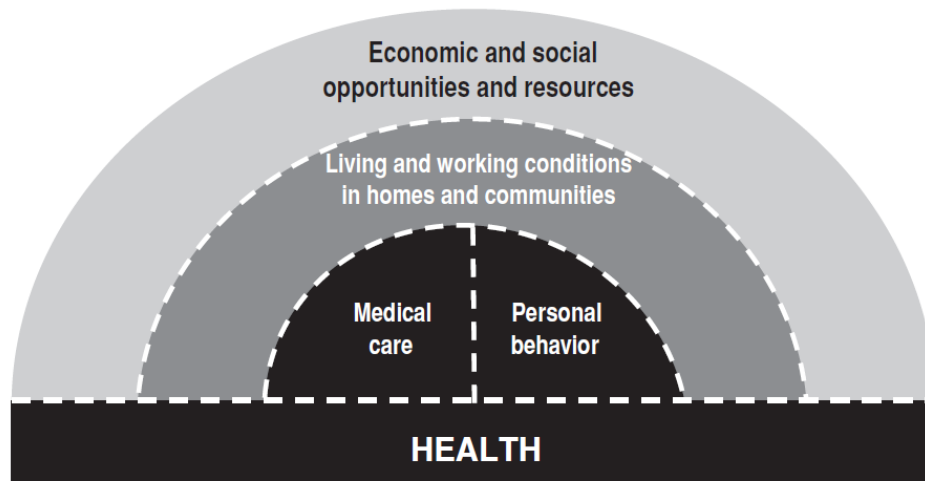


Figure 4 Upstream and Downstream Determinants Influencing Health.

*Note: Order Detail ID: 71974762 Annual review of public health by ANNUAL REVIEWS. Reproduced with permission of ANNUAL REVIEWS in the format Thesis/Dissertation via Copyright Clearance Center. Braveman, et al.⁸⁰

There are a number of upstream determinants that affect the health of individuals and populations. For example, research evidence shows that lower socioeconomic status (or more disadvantaged), is associated with poorer health, and higher educational attainment affects not only knowledge about best health practices, but also increases opportunities to acquire employment that offers higher wages and more benefits.⁸⁰ Higher educational attainment also makes unemployment, and its associated health consequences, less likely.⁸⁰ Other upstream determinants that affect health include race, ethnicity, and neighborhood conditions.⁸⁰ Social disadvantage may increase over time, and may also be passed on from generation to generation through multiple pathways.⁸⁰

Krieger discusses this accumulation of social disadvantage over time and across generations in respect to the importance of early life conditions, stating that individuals embody

cumulative exposures determined by their social context.⁸³ Each of the social factors is also experienced simultaneously with other social factors.⁸³ There is concern that those subpopulations most affected by social disadvantage may be the least likely (or able) to report it.⁸³ Krieger references the ecosocial inverse hazard law, which states that “the accumulation of health hazards tends to vary inversely with the power and resources of the populations affected.”^{83(p941)}

In addition to social and economic, physical and environmental (including living and working conditions), and personal determinants of health,⁸⁰ there are also several other determinants of health. There are political determinants of health, which include regulation of safety standards, and health services determinants of health, which include the access to and quality of available health services.⁸⁴ Additionally, there are biological and genetic determinants of health, which include factors such as age, sex, and the presence of inherited conditions.⁸⁴ All determinants of health interact with each other to affect the health of individuals and populations.⁸⁴

5.1.2 Disparities in Access to Healthcare and Genetic Services

Goldenberg, et al. examined potential barriers to integrating genetic services into healthcare for underserved populations in a 2013 editorial, where high cost, poor health insurance coverage, mistrust of healthcare professionals, and fear of discrimination were all listed as potential issues.⁵⁷ The authors discuss the potential assumption by professionals that underserved populations will not be interested in using genetic services as there are already difficulties in acquiring basic healthcare services, and that even if they are interested, they will not have the agency to use this knowledge to affect change.⁵⁷ The authors argue that this viewpoint is harmful in that it may act as a self-fulfilling prophecy, diminishing the future availability of genetic services

to underserved populations and increasing the socioeconomic divide between advantaged and disadvantaged peoples.⁵⁷

This inequity in access to genetic services was also discussed by Suther, et al., specifically regarding racial and ethnic disparities.⁵⁸ Barriers including distrust in the medical system in general and specifically concern over misuse of genetic testing, a lower level of knowledge regarding genetic testing, and a deficit of private health insurance coverage were found to be more prevalent in minority groups (Blacks and Latinos) when compared to non-Hispanic Whites.⁵⁸ The authors stated concern regarding these findings in that inequitable access to genetic services would widen the gap between different populations and increase health disparities.⁵⁸

Suther, et al. also completed a review of 18 research studies analyzing primary care physicians' perceived barriers to their patients accessing genetic services.⁵⁵ The four most common barriers identified were inadequate knowledge of basic genetics, lack of detailed or updated family histories, lack of confidence, and a lack of referral guidelines.⁵⁵ Delikurt et al., performed a review of nine studies in order to identify barriers to referral to genetic services in which both individual and institutional barriers were recognized.⁵⁴ Barriers at the individual level included a lack of awareness of personal risk and of medical history of family members, as well as a lack of knowledge of genetic services.⁵⁴ Institutional barriers, or those related to non-genetic healthcare professionals, included a lack of awareness of patient risk factors, obtaining adequate family history, knowledge on genetics, and awareness of genetic services, as well as inadequate coordination of referrals.⁵⁴ Additionally, a lack of a genetics workforce was also identified as an institutional barrier.⁵⁴ The findings of the two articles by Suther, et al.⁵⁵ and Delikurt, et al.⁵⁴ dovetail with each other, in that the barriers identified relating to non-genetic healthcare providers were consistent with each other.

In a research article by Beene-Harris, et al., which was included in the review by Delikurt, et al.,⁵⁴ specific individual and institutional barriers were identified during multiple focus group sessions.⁵ Here, individual barriers described were lack of: awareness of personal risk, knowledge of genetic services and resources, and trust.⁵ Institutional barriers identified were lack of knowledge of genetic services and workforce, as well as issues with coordination of care, cost/insurance, and location from services.⁵ The authors stated, “Many of the barriers identified by focus group participants correspond with the concept of access as defined by Penchansky and Thomas in 1981; in particular, access is conceptualized to consist of several dimensions: availability, accessibility, accommodation, affordability, and acceptability.”^{5(p17)} The current study aimed to analyze multiple factors that may impact the ability of underserved populations to access genetic services.

5.1.3 Clinic Directory

The New York Mid-Atlantic Consortium (NYMAC) works to connect individuals with genetic services, especially individuals who are underserved.²¹ This Regional Genetics Network (RGN) encompasses eight states: Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia.²⁰ NYMAC has several projects that work to achieve the goal of increasing access to genetic services, one of which was the creation of a genetic clinic services directory that lists all of the clinic sites within the NYMAC region. This directory was generated in house and includes specific properties of each clinic site, including their location; this directory will soon be incorporated into a new American College of Medical Genetics and Genomics (ACMG) genetic clinic searchable database to provide referral information for individuals to genetic clinics.⁸⁵

To increase access to genetic services, NYMAC set up a Genetic Services Referral Phone Line that is associated with the clinic directory.⁸⁶ Callers are asked to enter their zip code and then may ask questions such as those regarding the location of genetic clinics by specialty, what a genetic counseling appointment would entail, and obtaining more information about a specific genetic condition or concern.⁸⁶ Both patients and family members can call the phone line for this type of information. Individuals with questions about a specific genetic disease may be transferred to the Genetic and Rare Diseases Information Center (GARD) where information specialists will address their questions.⁸⁷ Individuals requesting support with healthcare, mental health care, educational, legal, or emotional support can also be transferred to the official family-led organization in their state that deals with the particular area of concern. Family-led organizations are those that are staffed by family members of individuals with special healthcare needs, or self-advocates with special healthcare needs, that help patients and family members with individual concerns regarding the topics above and provide training for advocacy work.³

Healthcare providers may also contact the Genetics Services Referral Phone Line to find out more about how to refer their patients to genetic services, as well as to ensure their clinic is listed in the directory. If the clinic has a particular specialty, such as prenatal genetic services, the specialty will also be listed in the directory. The clinic directory contains information on both physical main clinic and outreach sites as well as telegenetic clinic sites, which can help individuals connect with specialists that are not geographically nearby.

One of the barriers to access to healthcare services, including specialty services like genetic services, is the uneven geographical distribution of clinic sites.³³ There are 579 physical clinic sites in the NYMAC region listed in the clinic directory. A heat map showing the locations of these physical genetic clinics listed in the directory is shown by county in Figure 5. There are

large areas where there are no clinic sites, indicated by the grey color. In order to better understand barriers to access to genetic services specifically for the underserved subpopulation, an investigation into the demographics of underserved regions and whether or not there is a correlation between these demographics and the number of physical clinic sites per county was undertaken.

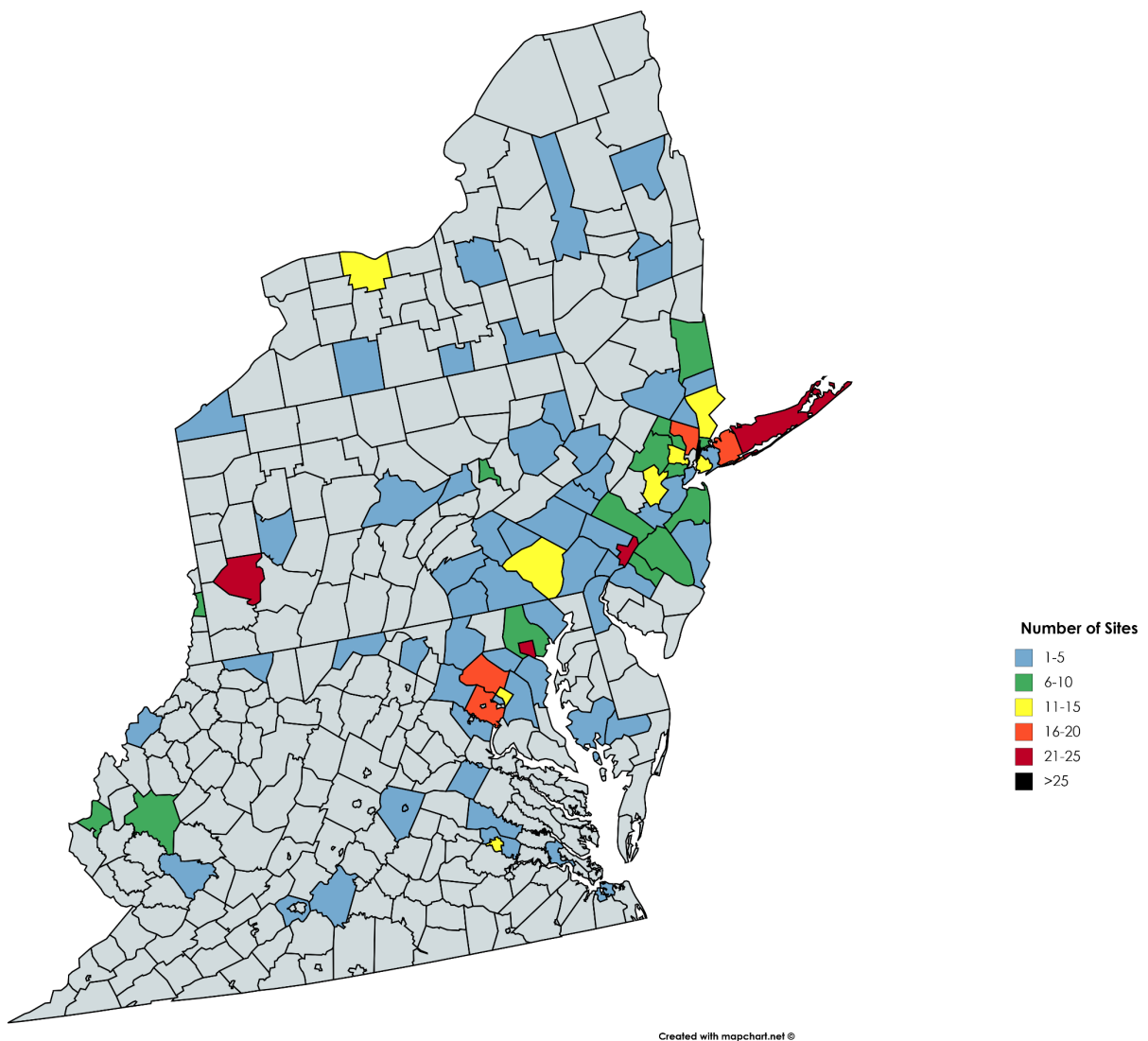


Figure 5 Color Coded Map: Number of Clinic Sites by County in the NYMAC region.

*Note: Made using United States Counties. MapChart.⁸⁸

5.1.4 HRSA MUA/P Designations

The NYMAC project to create a clinic directory and staff the Genetic Services Referral Phone Line was funded by the Genetic Services Branch in the Health Resources and Services Administration's (HRSA) Maternal and Child Health Bureau (MCHB). HRSA's mission is "to improve health outcomes and address health disparities through access to quality services, a skilled health workforce, and innovative, high-value programs."⁸⁹ In order to establish metrics to determine types of funding opportunities, the effects of programs, and the changes in underserved areas and populations, as well as other initiatives, HRSA has identified shortage areas.⁸² Medically Underserved Areas (MUA) and Medically Underserved Populations (MUP) are two different designations for shortage areas; when analyzed together they are referred to as MUA/P.⁹⁰ MUAs represent areas and MUPs represent populations that have too few primary care physicians (PCPs). MUPs may include populations with low socioeconomic status, as well as populations with cultural and linguistic diversities, which may act as barriers to healthcare.⁹⁰ This study used HRSA MUA/P designations to determine which counties were underserved for multiple analyses.

5.1.5 Purpose of Study

This study aimed to investigate the demographic differences between regions with different MUA/P designations in the NYMAC region. This study also aimed to determine if there is a correlation between each of these demographics and the number of physical clinic sites per county. This may help to better elucidate the barriers to accessing genetic services, the understanding of which would help in working to overcome these barriers.

5.1.5.1 Study Aims

This study has two aims.

Study Aim 1 To determine what the demographics are of the different categories of MUA/P regions. The three categories include 1) counties that are entirely underserved, 2) counties that are entirely not underserved, and 3) counties that are partially underserved.

Study Aim 2 To determine if there are correlations between specific demographics and the number of physical clinic sites in each category of county. If this is the case, then determine to what extent there is a correlation, and the direction of the correlation, between specific demographics and the number of physical clinic sites in each category of county.

5.2 Methods

5.2.1 Data Sources

All of the data pertaining to individual counties except for number of clinics per county and MUA/P designation were acquired from the County Health Rankings & Roadmaps, which is a Robert Wood Johnson Foundation program.⁸¹ The data regarding the MUA/P county designations were acquired through HRSA's Map Tool.⁹¹ The data regarding the number of clinics in each county was generated in house. This study was reviewed and determined to be not human

subjects research by the University of Pittsburgh Institutional Review Board, please see the letter in **Appendix E**. All descriptive statistical analyses were performed using IBM SPSS Statistics 25.

The clinic directory was used as the source of the data on the number of clinic sites per county. This directory was made in house by calling or emailing each clinic to confirm accuracy of information. For those few clinics that could not be contacted directly, the clinic site information was fact checked through their websites. The HRSA Map Tool was used to check each county in the NYMAC region individually to determine its MUA/P designation.⁹¹ The counties were placed into one of three designations: completely underserved county, completely not underserved county, and partially underserved county. The County Health Rankings and Roadmaps was used to determine all other county data, including rural percentage, population, income ratio, median household income, PCP rate, percent uninsured, percent with some college, and percent unemployed.⁸¹ The County Health Rankings program is a collaboration between the Robert Wood Johnson Foundation, the largest philanthropic organization in the country focused on improving health, and the University of Wisconsin Population Health Institute.⁹²

5.2.2 Specific Demographics Investigated

The demographics investigated for each county included rural percentage, population, income ratio, median household income, PCP rate, percent uninsured, percent with some college, and percent unemployed. All of these data were acquired from the County Health Rankings and Roadmaps.⁸¹ The rural percentage and the population measurements for each county were taken from the 2010 and 2016 Census Population Estimates, respectively.⁹³ The percent of individuals in the county who were uninsured data was generated from the Small Area Health Insurance Estimates of 2015.⁹³ The percent of individuals in the county with some college education data

was generated from the 2012-2016 American Community Survey.⁹³ The percent of individuals in the county who were unemployed data was generated from the Bureau of Labor Statistics of 2016.⁹³ The median household income data was taken from the Small Area Income and Poverty Estimates of 2016.⁹³ The income ratio data were also generated from the 2012-2016 American Community Survey.⁹³ The income ratio represents the ratio of household income at the 80th percentile to the household income at the 20th percentile,⁸¹ giving a measurement of the discrepancy of household income per county. The PCP rate data was supplied by the Area Health Resource File/American Medical Association of 2015.⁹³ The PCP rate represents the number of primary care physicians per 100,000 individuals in the population of that county.⁸¹

5.2.3 Descriptive Analyses

A histogram was generated for each of the variables, showing the distribution of values for data points in all counties. The histogram showing the number of clinic sites per county is presented in Figure 6 below. The data point with the highest number of clinic sites is for New York County in New York, NY, where there are 53 clinic sites present. This data point was considered an outlier, as the next closest data point is Baltimore City County in Baltimore, MD, with 25 clinic sites.

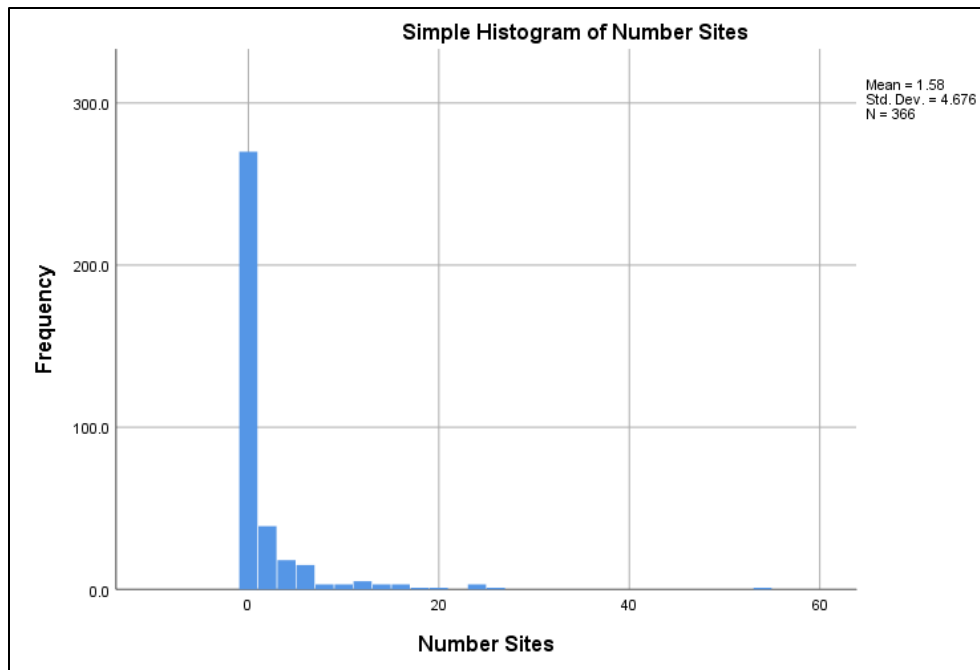


Figure 6 Histogram of Number of Clinic Sites per County, Including New York County.

The same histogram is shown below, with the outlier of New York County removed. All data were analyzed using the data set without New York County.

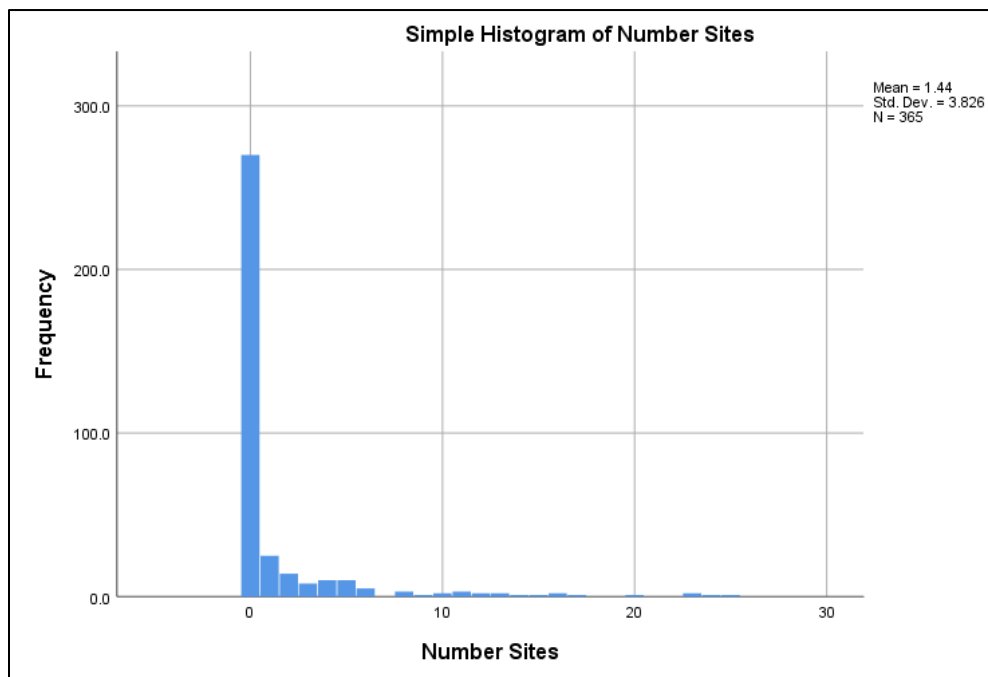


Figure 7 Histogram of Number of Clinic Sites per County, Not Including New York County.

The demographic variable of rural percentage for each county was investigated for use in further statistical analyses; however, the histogram, which is shown below, indicated a non-normal distribution, a normal distribution being the shape of a bell curve. This type of distribution is not compatible with the statistical analyses used to analyze the other variables, and so it was discarded as an option.

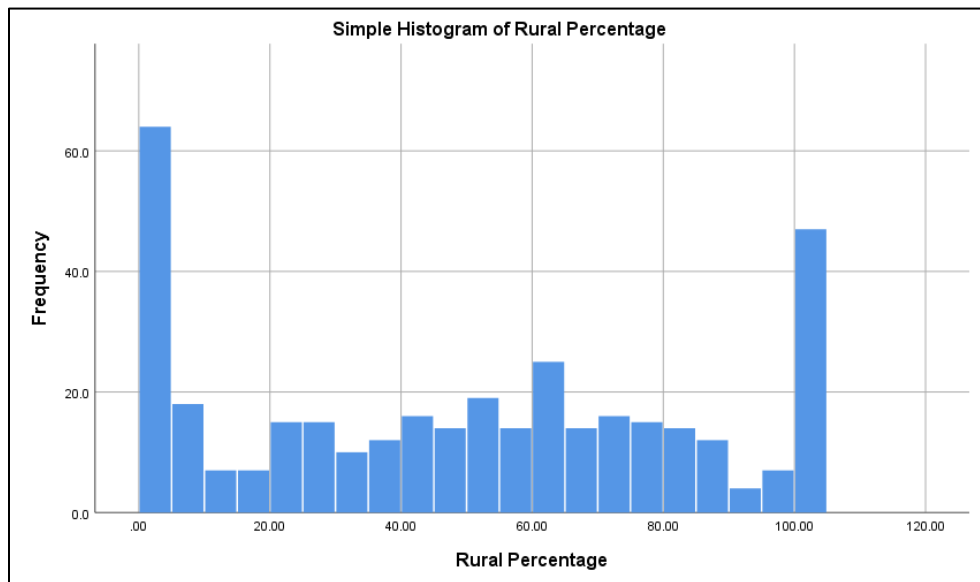


Figure 8 Histogram of Rural Percentage by County.

5.2.4 Statistical Analyses

A one-way ANOVA could not be run in a standard way as the variances between the variables were not considered equal. The significance values of the Levene Statistics for the test of homogeneity of variances indicated a significant difference in the variances in the following variables: population, median household income, PCP rate, percent uninsured, percent unemployed, and the number of clinic sites per county. Two variables did not have a significant

difference between their variances: income ratio, and percent some with college. A standard one-way ANOVA can be run only if all the variables have equal variances. As such, a Welch's ANOVA was performed. A Games-Howell post-hoc test was performed, instead of Tukey, also in order to account for the unequal variances.

A box and whiskers plot was generated for each variable, showing the difference in the distributions of each variable between the different MUA/P designations. This is a visualization of the data analyzed in the Welch's ANOVA and the Games-Howell post-hoc test.

A correlation analysis was also run between the number of clinic sites per county and each of the demographic variables: population, income ratio, median household income, PCP rate, percent uninsured, percent with some college, and percent unemployed. If a statistically significant Pearson correlation r value is obtained it would indicate that the correlation between the number of clinics per county and the demographic variable being analyzed is not likely due to chance.

A scatter plot with a best fit line was generated for each of the variables analyzed. This is a visualization of the data analyzed in the correlation analysis. The r^2 value represents the goodness of fit of the line with the data points.

5.3 Results

5.3.1 Descriptive Analyses

Descriptive analyses were used to examine the quality of the data for use in further statistical analyses. The number of data points, the average value, and the standard deviation are

given for all variables: population, income ratio, median household income, PCP rate, percent uninsured, percent some college, percent unemployed, and the number of clinic sites. Each set of data is additionally broken down by the MUA/P designation the county was classified as: underserved, not underserved, or partially underserved. These data are shown in Table 6. These data are consistent with the social determinants of health and illustrate disparities in multiple upstream factors relating to education, income (and other employment variables), and health.⁸⁰ Underserved areas show a lower median household income, PCP rate, percent with some college, and number of clinic sites; underserved areas also show a higher percent uninsured and percent unemployed.

Table 6 Descriptives of Variables by MUA/P Designation.

| | Region Type | N | Mean | Std. Deviation |
|----------------------------------|-----------------------|----------|-------------|-----------------------|
| Population | Underserved | 161 | 33405.77 | 37114.53 |
| | Not Underserved | 34 | 85359.56 | 100083.67 |
| | Partially Underserved | 170 | 290836.99 | 397578.50 |
| | Total | 365 | 158144.78 | 300836.91 |
| Income Ratio | Underserved | 161 | 4.50 | .61 |
| | Not Underserved | 34 | 4.23 | .54 |
| | Partially Underserved | 170 | 4.57 | .74 |
| | Total | 365 | 4.51 | .68 |
| Median Household Income | Underserved | 161 | 47998.19 | 13423.51 |
| | Not Underserved | 34 | 65174.62 | 23461.55 |
| | Partially Underserved | 170 | 60226.66 | 17876.70 |
| | Total | 365 | 55293.64 | 17930.65 |
| PCP Rate | Underserved | 153 | 49.92 | 33.01 |
| | Not Underserved | 33 | 91.81 | 96.33 |
| | Partially Underserved | 170 | 72.30 | 34.23 |
| | Total | 356 | 64.49 | 45.29 |
| Percent Uninsured | Underserved | 161 | 10.30 | 3.11 |
| | Not Underserved | 34 | 7.99 | 2.88 |
| | Partially Underserved | 170 | 8.09 | 2.22 |
| | Total | 365 | 9.06 | 2.92 |
| Percent with Some College | Underserved | 161 | 52.23 | 9.91 |
| | Not Underserved | 34 | 63.94 | 11.50 |
| | Partially Underserved | 170 | 62.63 | 9.77 |
| | Total | 365 | 58.16 | 11.29 |
| Percent Unemployed | Underserved | 161 | 5.71 | 1.94 |
| | Not Underserved | 34 | 4.43 | 1.00 |
| | Partially Underserved | 170 | 5.26 | 1.22 |
| | Total | 365 | 5.38 | 1.61 |
| Number of Sites | Underserved | 161 | .07 | .41 |
| | Not Underserved | 34 | 1.24 | 2.66 |
| | Partially Underserved | 170 | 2.78 | 5.14 |
| | Total | 365 | 1.44 | 3.83 |

Histograms for each of the demographic variables, including population, income ratio, median household income, PCP rate, percent uninsured, percent with some college, and percent unemployed, were generated and indicated compatibility with planned statistical analyses. These histograms are shown in **Appendix F**.

5.3.2 Statistical Analyses: Demographics of MUA/P Designated Regions

The demographics of the regions in each of the three MUA/P designations were visualized using box and whiskers plots, where each distribution is represented vertically. Additionally, the number of clinics in each county was plotted in this way to show the variation between MUA/P designations. These plots are shown in Figure 9 below. The findings are again consistent with the social determinants of health, in that there are fewer social and economic opportunities and resources, or fewer advantageous characteristics of upstream factors,⁸⁰ in the underserved regions than in the not underserved regions. Additionally, while the median household income is lower in underserved areas, the income ratio is higher in underserved areas. This finding was further investigated with regards to the number of clinic sites per county, which is described below.

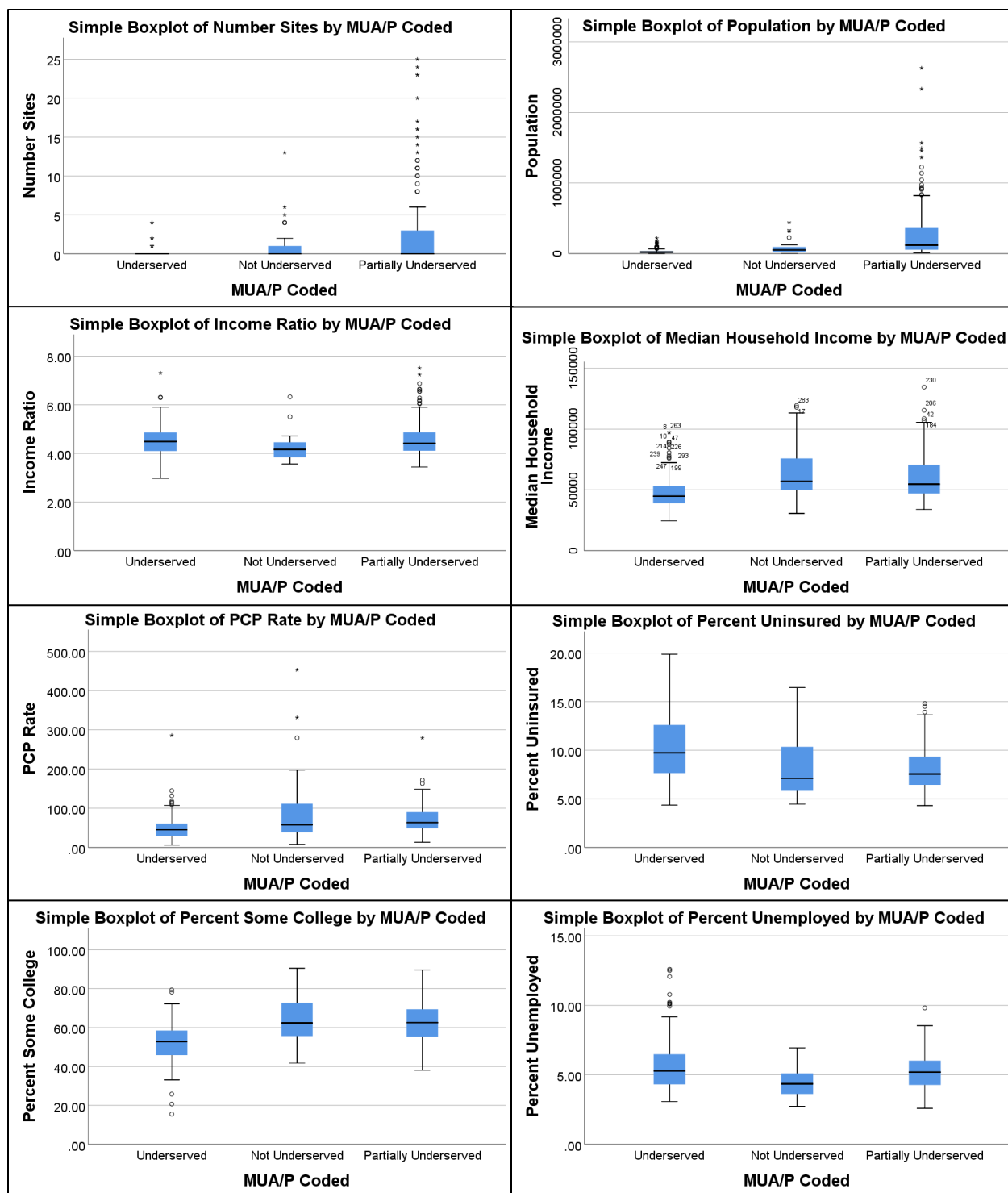


Figure 9 Box and Whisker Plots of MUA/P Coded Regions and Number of Clinic Sites per County, Population, Income Ratio, Median Household Income, PCP Rate, Percent Uninsured, Percent with Some College, and Percent Unemployed.

Statistical analyses were run to determine if the demographics and number of clinics in each of the MUA/P designation were significantly different from one another. A Welch's ANOVA was run to determine if there was any significant difference between any of the MUA/P designations across the variables listed above, the results of which are shown in Table 7. Each demographic tested, as well as the number of clinics, generated a p value of less than .001, except income ratio, which generated a p value of less than .05, indicating there was significant difference between groups for all of the variables, and that post hoc testing was warranted.

Table 7 Welch ANOVA Results.

| | Analysis Type | df | F | Sig. |
|--------------------------------|----------------------|-----------|----------|-------------|
| Population | Between Groups | 2 | 37.697 | <.001 |
| | Within Groups | 362 | | |
| | Total | 364 | | |
| Income Ratio | Between Groups | 2 | 3.532 | .030 |
| | Within Groups | 362 | | |
| | Total | 364 | | |
| Median Household Income | Between Groups | 2 | 28.718 | <.001 |
| | Within Groups | 362 | | |
| | Total | 364 | | |
| PCP Rate | Between Groups | 2 | 18.028 | <.001 |
| | Within Groups | 353 | | |
| | Total | 355 | | |
| Percent Uninsured | Between Groups | 2 | 30.480 | <.001 |
| | Within Groups | 362 | | |
| | Total | 364 | | |
| Percent Some College | Between Groups | 2 | 50.943 | <.001 |
| | Within Groups | 362 | | |
| | Total | 364 | | |
| Percent Unemployed | Between Groups | 2 | 10.379 | <.001 |
| | Within Groups | 362 | | |
| | Total | 364 | | |
| Number Sites | Between Groups | 2 | 23.431 | <.001 |
| | Within Groups | 362 | | |
| | Total | 364 | | |

A Games-Howell post hoc test was performed to determine which comparisons between groups were statistically significant. As there were three MUA/P designations, three tests were run for each variable, to test all possible combinations for significant differences. The results of this analysis are shown in Table 8.

Table 8 Games-Howell Multiple Comparisons Results.

| | Region Type 1 | Region Type 2 | Sig. |
|----------------------------------|-----------------------|-----------------------|-------------|
| Population | Underserved | Not Underserved | .014* |
| | Partially Underserved | Underserved | <.001** |
| | Not Underserved | Partially Underserved | <.001** |
| Income Ratio | Underserved | Not Underserved | .032* |
| | Partially Underserved | Underserved | .658 |
| | Not Underserved | Partially Underserved | .008** |
| Median Household Income | Underserved | Not Underserved | .001** |
| | Partially Underserved | Underserved | <.001** |
| | Not Underserved | Partially Underserved | .481 |
| PCP Rate | Underserved | Not Underserved | .048* |
| | Partially Underserved | Underserved | <.001** |
| | Not Underserved | Partially Underserved | .491 |
| Percent Uninsured | Underserved | Not Underserved | <.001** |
| | Partially Underserved | Underserved | <.001** |
| | Not Underserved | Partially Underserved | .981 |
| Percent with Some College | Underserved | Not Underserved | <.001** |
| | Partially Underserved | Underserved | <.001** |
| | Not Underserved | Partially Underserved | .811 |
| Percent Unemployed | Underserved | Not Underserved | <.001** |
| | Partially Underserved | Underserved | .032* |
| | Not Underserved | Partially Underserved | <.001** |
| Number Sites | Underserved | Not Underserved | .040* |
| | Partially Underserved | Underserved | <.001** |
| | Not Underserved | Partially Underserved | .032* |

Note: ** Correlation is significant to the 0.01 level and * Correlation is significant to the 0.05 level

All results marked with asterisks were statistically significant, indicating the difference in the variables tested between that pair of MUA/P designations were unlikely to be due to chance alone.

5.3.3 Statistical Analyses: Correlation between Demographics and Clinics in Counties

Correlations were run between each of the six independent variables and the number of clinic sites in each county. All Pearson correlations (r values) returned were significant, as shown in the row labeled Sig. (p values) in the table below, indicating that the correlation between each of the independent variables and the numbers of clinic sites in each county was unlikely to be due to chance alone.

Table 9 Correlations between Demographic Variables and Number of Clinic Sites per County.

| Number of Sites | | Population | Income Ratio | Median Household Income | PCP Rate | Percent Un-insured | Percent Some College | Percent Un-employed |
|-----------------|---------------------|------------|--------------|-------------------------|----------|--------------------|----------------------|---------------------|
| | Pearson Correlation | .759** | .279** | .308** | .285** | -.106* | .341** | -.130* |
| | Sig. (2-tailed) | <.001 | <.001 | <.001 | <.001 | .043 | <.001 | .013 |
| | N | 365 | 365 | 365 | 356 | 365 | 365 | 365 |

*Note: ** Correlation is significant to the 0.01 level. * Correlation is significant to the 0.05 level.

The demographic variable of population had the largest positive r value, indicating that the strongest direct correlation was between an increase in the population of a county and an increase in the number of clinic sites in that county. This indicates that the more populous a county, the more likely it is to have a larger number of physical clinic sites. This data matches the previously shown heat map, where counties in heavily populated cities have a larger number of physical clinic sites present. The two demographic variables with negative r values were percent uninsured and percent unemployed, which show an inverse relationship between the number of clinic sites in a county and these two demographic variables. As these r values are the smallest magnitude of all the r values calculated, these two demographic variables have the weakest correlation with the number of clinic sites in the county; however, they both have statistically significant p values,

indicating that the correlations are significant. The pattern of fewer clinic sites in counties with a higher percent of both uninsured and unemployed individuals is consistent with the social determinants of health, which illustrates the link between social inequities and poorer health outcomes.

Here, both the income ratio and the median household income have a positive correlation, indicating they both increase with an increasing number of clinic sites per county. These data are shown in Table 9 and are further described in the discussion section. Scatter plots visualizing the correlation for each of the independent variables and the number of clinic sites in each county are shown in **Appendix G**.

5.4 Discussion

5.4.1 Data Analyses

A Welch ANOVA with a Games-Howell post-hoc test was done in order to see if the demographics between the three MUA/P designations were significantly different (Tables 7 and 8). To further visualize these results, Table 10 shows each of the variables is listed in the first column with the three types of MUA/P designations listed to the right of each variable. The first row is always labeled with the underserved designation, the second row with the not underserved designation, and the third row with the partially underserved designation. If a designated region is not in the same column as another designated region, that indicates that there is a significant difference between the designated regions being compared. If a designated region is in the same column as another designated region, that indicates that there is not a significant difference

between the designated regions being compared. For example, when comparing populations across the three types of MUA/P regions, each is in its own column, as each designated region is significantly different from both other designated regions.

Table 10 Variables across MUA/P Designations.

| | | | |
|----------------------------------|-----------------------|-----------------------|-----------------------|
| Population | Underserved | | |
| | | Not Underserved | |
| | | | Partially Underserved |
| Income Ratio | Underserved | | |
| | | Not Underserved | |
| | Partially Underserved | | |
| Median Household Income | Underserved | | |
| | | Not Underserved | |
| | | Partially Underserved | |
| PCP Rate | Underserved | | |
| | | Not Underserved | |
| | | Partially Underserved | |
| Percent Uninsured | Underserved | | |
| | | Not Underserved | |
| | | Partially Underserved | |
| Percent with Some College | Underserved | | |
| | | Not Underserved | |
| | | Partially Underserved | |
| Percent Unemployed | Underserved | | |
| | | Not Underserved | |
| | | | Partially Underserved |
| Number of Clinic Sites | Underserved | | |
| | | Not Underserved | |
| | | | Partially Underserved |

These data indicate that almost all demographics differ significantly between each of the MUA/P designation types, which will be discussed further below. There are five exceptions, where the variable is not significantly different between the MUA/P designations. These include 1) income ratio between the underserved and partially underserved regions, 2) median household income between the not underserved and partially underserved regions, 3) PCP rate between the

not underserved and partially underserved regions, 4) percent uninsured between the not underserved and partially underserved regions, and 5) percent with some college between the not underserved and partially underserved regions.

Where the variables differ between specific MUA/P designations, this indicates that the underserved, not underserved, and partially underserved regions are distinct in the majority of demographic variables tested. If only underserved and not underserved regions are directly compared, there is a significant difference in all demographics including population, income ratio, median household income, PCP rate, percent uninsured, percent with some college, and percent unemployed between these two regions. There is also a significant difference in the number of clinic sites between underserved and not underserved regions with underserved regions having fewer clinic sites. The data show that when compared to the not underserved regions, the underserved regions have lower population, higher income ratio, lower household income, lower PCP rate, higher percent uninsured, lower percent with some college, higher percent unemployed, and lower number of clinic sites. The lower PCP rate in the underserved regions indicates that these areas have fewer primary care physicians for the population than not underserved areas.

When analyzing these data in light of the social determinants of health, they are consistent with the population in underserved regions having less access to economic and social opportunities and resources, specifically relating to income and education, than the population in the not underserved regions.⁸⁰ Braveman, et al. states, “medical care alone cannot adequately improve health overall or reduce health disparities without also addressing where and how people live.”^{80(p381)} These data directly measure where and how people live, completing the first step in the long process of addressing social determinants of health and reducing health disparities. These data may be able to help RGNs and the NCC better reach underserved subpopulations.

The greater the discrepancy between the larger household incomes (at the 80th percentile) and the lower household incomes (at the 20th percentile) in the county results in the larger the income ratio. The higher income ratio in the underserved regions indicates that there is a larger household income discrepancy in these areas than in the not underserved areas.

Regarding partially underserved regions, the percentage of each county that is underserved versus not underserved is not known, and so for any given variable, the partially underserved regions may be more similar to underserved regions, or to not underserved regions. The five comparisons that returned results that were not statistically significant were all between a partially underserved region and either an underserved or not underserved region. This indicates that for five of the seven demographic variables, the partially underserved areas were not statistically significantly different from one of either the underserved or not underserved areas. More information regarding these partially underserved counties would be needed to achieve more specific results in statistical analyses.

A correlation analysis was also performed between the number of clinic sites and each of the demographic variables. All correlations returned significant results, indicating that a correlation exists between the number of clinic sites and each of the demographic variables, and that the correlation is unlikely to be due to chance.

The population, income ratio, median household income, PCP rate, and percent with some college all have positive correlations with the number of clinic sites, indicating that as each of the demographic values increase, so does the number of clinic sites in that county. The percent uninsured and percent unemployed both have negative correlations with the number of clinic sites, indicating that as each of these demographic values increase, the number of clinic sites in that county decrease. This indicates that there are disparities in genetic services, where those with less

social advantage live in areas where there are fewer genetic clinic sites, a cyclical problem where each characteristic reinforces the other. All of these correlation patterns are consistent with the findings from the demographic studies of the underserved and not underserved regions, and thus the social determinants of health previously described, except for the income ratio finding.

The correlation shows that as the income ratio increases (the discrepancy between high and low household incomes), so too does the number of clinic sites in that county. Previous results in these statistical analyses indicate that income ratio is higher in underserved areas. These two findings would imply that there are a higher number of clinic sites in underserved areas; however, it was found that there are a lower clinics in underserved areas. It is possible that in the areas with higher income ratios that those individuals at the upper end of the income range are both the driving force behind the higher number of physical clinic sites as well as the individuals in the county able to utilize the clinic sites.

These data can be further investigated by analyzing the trends in the median household income and number of clinic sites between underserved and not underserved MUA/P designated regions. The underserved regions have a lower median household income, and median household income increases with increasing number of clinic sites per county. These are both consistent with the social determinants of health and a straightforward relationship of having more clinic sites in not underserved areas. The relationships with income ratio are not as straightforward.

In underserved areas where there is a higher income ratio, the median household income is lower. This indicates that there is less income overall in these underserved areas, which may increase income discrepancy and result in a larger income ratio. In regions with more clinic sites per county where there is a higher income ratio, the median household income is higher. This indicates that there is more income overall in these regions with more clinic sites, which may also

increase income discrepancy and result in a larger income ratio. These findings would support the inference that individuals with a higher median household income are a driving force behind the presence of more physical clinic sites in that county. This speculation would need to be investigated further to determine the cause of the positive correlation between the income ratio and the number of clinic sites per county.

When assessing the correlation between the number of clinic sites and the demographic variables, the higher the magnitude of the Pearson correlation value, the stronger the correlation between the two variables being investigated. Here, the number of clinic sites is correlated in decreasing order of strength with the following demographic variables: population, percent with some college, median household income, PCP rate, income ratio, percent unemployed, and percent uninsured. This would indicate that those demographic variables with stronger correlations (at the beginning of the list) would be more influential in affecting the number of clinic sites in that county, or conversely that the number of clinic sites in that county is more influential on those demographic variables (at the beginning of the list) of the individuals who live in that county. While the strength of each correlation is given in this analysis, it is likely that all of the demographic variables have an effect on each other. This is supported by Krieger's discussion of the accumulation of concurrent social disadvantages.⁸³ Additionally, correlation does not equal causation. There may be other factors that affect both the demographic variable of interest and the number of clinic sites in the county, which would not be shown in this analysis. There may also be self-selection occurring in many of the counties, where areas that are not underserved and/or have more clinic sites draw in individuals that have certain demographic characteristics.

5.4.2 Guiding Efforts to Address Gaps in Access to Genetic Services

This study supports the theory of a significant difference in demographics between different MUA/P designations of underserved and not underserved areas, as well as correlations between certain demographic characteristics and the number of genetic clinic sites in that county. It also connects the social determinants of health with the proximity to genetic services and confirms that certain areas are underserved with respect to healthcare, and specifically genetic services. By focusing future efforts on areas designated as underserved and counties with fewer genetic clinic sites, this study may help to address gaps in genetic services. An initial effort may focus on expanding services in areas without genetic clinic sites and areas that are designated as underserved. Additionally, if particular demographic characteristics are confirmed to be predictive of poorer access to genetic services and areas that are underserved, future efforts could be focused where these particular demographic characteristics are prevalent. This study may also justify funding on projects addressing these issues.

Partnering with family-led organizations or parent mentors and Regional Genetic Networks going forward may help people to better access genetic services, especially in underserved areas. Additionally, the increased incorporation of the medical home model, which involves care coordination and family-centered care may also help individuals to better access genetic services.

5.4.3 Limitations

In this study there was difficulty statistically differentiating between the MUA/P designated counties that were partially underserved from either underserved or not underserved. While the partially underserved counties could be distinguished from one of either the underserved

or not underserved counties, they could not be distinguished from both in all analyses. Additionally, there were fewer counties designated as not underserved than either underserved or partially underserved. It may be possible to statistically differentiate between the underserved areas and not underserved areas from the partially underserved areas if there were more data points, possibly achieved by expanding the area investigated beyond the NYMAC region. There are also limitations to secondary data analysis, as the County Health Rankings data was not originally collected for the purpose of this study. Lastly, this was a cross-sectional study, investigating the variables of different counties at a point in time, and therefore can interpret correlation but not causation.

5.4.4 Future Directions

By gathering more information about partially underserved counties, more significant results may be found in the statistical analyses. This could be done by using a percentage of the area or population in that county that is considered underserved versus not underserved. It is also possible to break down areas into finer measurements than county, possibly by using zip code or by the MUA/P service area name or identification number. Each county has multiple zip codes and MUA/P service areas within it, and these finer area measurements do not necessarily line up with county lines. Analyzing more information regarding the demographics of these counties may also give insight into why the income ratio increases as the number of clinic sites in the area increases. Future studies could include the analysis of clinic site specialty and telegenetics presence to determine if there are patterns with these properties as well. This cross-sectional study could interpret correlation, but not causation. It is possible that a longitudinal study of these areas over time may indicate the cause of the relationships between variables. Causation may be found

to be due to another variable altogether, and causation may also be linked between demographic variables that affect one another.

5.5 Conclusions

Descriptive and statistical analyses of demographic, MUA/P designation, and clinic site data allowed for multiple assessments, and showed that the demographics between the underserved and not underserved areas in the NYMAC region were significantly different for all variables investigated. The data show that the underserved regions have a lower population, a higher income ratio, a lower median household income, a lower PCP rate, a higher percent uninsured, a lower percent with some college, a higher percent unemployed, and a lower number of clinic sites. The higher income ratio, lower median household income, lack of primary care providers and clinic sites, lower percent with some college, and higher percent of both uninsured and unemployed individuals in the underserved regions is consistent with the social determinants of health and a lack of opportunity and resources. More data are needed to be able to draw significant conclusions about counties that are a combination of underserved and not underserved areas and populations.

A correlation was also found between the number of clinic sites and each demographic variable investigated. These data are again consistent with the social determinants of health, where socioeconomic status strongly influences the health of populations. The income ratio of the county was also directly correlated with the number of clinic sites in the county, a finding that merits further investigation. More insight into the characteristics of the partially underserved counties would generate a more specific analysis of the demographics within them. An understanding of the strength of the correlations of demographic variables with the number of clinic sites in the

county, with each other, and with other outside factors would also benefit from further investigation. Longitudinal studies may elucidate causation in these correlations.

The findings in this study support the theory of a significant difference in demographic variables between areas designated as underserved and not underserved and a correlation of these demographic variables with the number of genetic clinic sites per county. This study links the number of genetic clinic sites per county directly with aspects of the social determinants of health. These findings may guide future efforts to improve access to genetic services, as well as justify the funding of projects addressing these issues.

Appendix A Institutional Review Board Approval

University of Pittsburgh Institutional Review Board

Human Research Protection Office
3500 Fifth Avenue, Suite 106
Pittsburgh, PA 15213
Tel (412) 383-1480
www.hrpo@pitt.edu

APPROVAL OF SUBMISSION (Expedited)

| | |
|----------|---|
| IRB: | STUDY19050146 |
| PI: | Sarah Brunker |
| Title: | Personal Experiences of Leaders of Family-Led Organizations in the NYMAC Region and their Role as Advocates |
| Funding: | None |
| Date: | May 28, 2019 |

On 5/28/2019, the Institutional Review Board reviewed and approved the above referenced application through the administrative review process. The study may begin as outlined in the University of Pittsburgh approved application and documents.

Approval Documentation

| | |
|---------------------|---|
| Review type: | Initial Study |
| Risk Level: | No greater than minimal risk |
| Approval Date: | 5/28/2019 |
| Expedited Category: | (6) Voice, video, digital, or image recordings, (7)(b) Social science methods |
| Determinations: | • Waiver of consent documentation |
| Approved Documents: | • InterviewQuestions.docx • ConsentScriptFinal_SEBEdits.docx • EmailtoLeaders.pdf |

As the Principal Investigator, you are responsible for the conduct of the research and to ensure accurate documentation, protocol compliance, reporting of possibly study-related adverse events and unanticipated problems involving risk to participants or others. The HRPO Reportable Events policy, Chapter 17, is available at <http://www.hrpo.pitt.edu/>.

Continuing review (CR) can be submitted by clicking "Create Modification/CR" from the active study at least 5 weeks prior to the expiration date.

Research being conducted in an UPMC facility cannot begin until fiscal approval is received from the UPMC Office of Sponsored Programs and Research Support (OSPARS). Contact OSPARS@upmc.edu with questions.

If you have any questions, please contact the University of Pittsburgh IRB Coordinator, [Teresa McKaveney](#).

Please take a moment to complete our [Satisfaction Survey](#) as we appreciate your feedback.

Appendix B Semi-Structured Interview Guide

Can you tell me about your background?

Can you tell me about the organization for which you work?

How did the organization come to be?

How has the organization evolved over time?

Can you tell me about your participation in the organization over time?

Can you tell me about your story as a family member of a person with a genetic condition?

What did you do when you were concerned about a genetic condition in one of your family members?

What has been your experience with genetic services/genetic counseling?

What do you think about genetic services/genetic counseling?

Have you been able to access genetic services/genetic counseling?

How do you think genetic services/genetic counseling have helped you and your family lead a healthier life?

What have people in your community/organization shared with you about their experiences with genetic services/genetic counseling?

Have they been able to access genetic services/genetic counseling?

What would you suggest for others in a similar position as you?

Anything else we should discuss that I missed?

Appendix C Email Contact to Leaders

Dear [insert name of family-led organization leader],

I am a dual degree student in the MS in Genetic Counseling and MPH in Human Genetics program at the University of Pittsburgh. I am also a student worker for NYMAC. For the completion of my thesis, I am interviewing leaders of Family-Led Organizations in the NYMAC region. Specifically, I am hoping to interview leaders of Parent-to-Parent organizations, Family-to-Family organizations, and State Affiliate Organizations of Family Voices in Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia. These interviews will be recorded and transcribed, and data will be analyzed qualitatively, specifically using thematic analysis.

The overarching goal of this study is to record and analyze interviews from leaders of family-led organizations where they represent themselves directly. This study seeks to explore the barriers addressed by individuals who are both family members of CYSHCN and leaders of family-led organizations; these barriers may also be faced by the general population. In focusing on the leaders' stories, challenges, outcomes, goals, and perceptions, the aim of this study is to generate a multi-dimensional picture of the experiences of family members of children and youth with special health care needs. This will include the leaders' personal experiences with genetic services and their roles in assisting members of their organization in obtaining services, which will allow for information to be gathered about access to genetic services within a larger context. This material may be used in the future to increase the understanding about, and improve access to, genetic services.

If you would like any more information on my background or my thesis project please feel free to contact me with any questions or comments. I look forward to hearing from you regarding your interest in participating in an interview. Thank you for all that you do!

Sincerely,

Sarah Brunker, PhD

[phone number] (mobile)

Appendix D Consent Script

This script documents consent to act as a participant in the research study: Personal Experiences of Leaders of Family-Led Organizations in the NYMAC Region and their Role as Advocates.

If you have any questions about your rights as a research subject or wish to talk to someone other than the research team, please call the University of Pittsburgh Human Subjects Protection Advocate toll free at 866-212-2668. You can contact the study investigator if you have any questions about the study, concerns, or complaints. Contact the principal investigator, myself, Dr. Sarah Brunker, at [email address], or the faculty mentor, Dr. Andrea Durst, at [email address].

You are being asked to take part in a research study. Research studies include only people who choose to take part. I will explain the study to you and will answer any questions you might have. You should take your time to make your decision.

The overarching goal of this study is to record and analyze interviews from leaders of Family-Led Organizations where they represent themselves directly. This study seeks to explore the barriers addressed by individuals who are both family members of children and youth with special health care needs (CYSHCN) and leaders of Family-Led Organizations; these barriers may also be faced by the general population. In focusing on the leaders' stories, challenges, outcomes, goals, and perceptions, the aim of this study is to generate a multi-dimensional picture of the experiences of family members of CYSHCN. This will include the leaders' personal experiences with genetic services and their roles in assisting members of their organization in obtaining services, which will allow for information to be gathered about access to genetic services within a

larger context. This material may be used in the future to increase the understanding about, and improve access to, genetic services.

I am interviewing leaders of Family-Led Organizations, specifically Parent-to-Parent organizations, Family-to-Family organizations, and State Affiliate Organizations of Family Voices in the NYMAC region, including Delaware, District of Columbia, Maryland, New Jersey, New York, Pennsylvania, Virginia, and West Virginia. These interviews will be approximately 60 minutes in length. I will conduct the interview in a private office. The interviews will be recorded and transcribed by myself. Participants may be asked to complete follow-up interviews lasting approximately 30 minutes in length. Data will be analyzed qualitatively, specifically using thematic analysis. Approximately 10-15 participants will be asked to enroll in this study. Data will be summarized into my master's thesis and submitted as a manuscript to an appropriate professional journal. These research activities are to be completed for research purposes only.

This study involves the potential risks of a breach of confidentiality of data collected during the interview process. Such risks to confidentiality will be minimized by limiting access to information collected to only Sarah Brunker and Andrea Durst, and storing identifiable and coded, or deidentified, data files on a departmental server and university cloud account. These locations have access restricted to specific personnel and are also password protected. Authorized representatives from the University of Pittsburgh Research Conduct and Compliance Office may review your data solely for the purpose of monitoring the conduct of this study. Per University of Pittsburgh policy all research records must be maintained for at least 7 years following final reporting or publication of a project. Only de-identified transcripts will be printed to assist with completion of qualitative analysis; these printouts will be shredded afterwards. We will make reasonable efforts to protect the privacy of your information. Although every reasonable effort

has been taken, confidentiality during Internet communication activities cannot be guaranteed and it is possible that additional information beyond that collected for research purposes may be captured and used by others not associated with this study.

Participants in this research study may experience personal benefits associated with having the opportunity to share their stories, challenges, and goals in a confidential environment. It is possible that there will be no direct benefit to you from participating in the study. However, this study may help healthcare professionals learn more about access to healthcare, and specifically, genetic services.

You can, at any time, withdraw from this research study. To formally withdraw your consent for participation in this research study you should provide a written and dated notice of this decision to the principal investigator of this research study, Dr. Sarah Brunker, at the address listed above. Your decision to withdraw from this study will have no effect on your current or future relationship with the University of Pittsburgh. It is possible that you may be removed from the research study by the researchers, if for example, it is necessitated by the research timeline.

If a participant withdraws from the research before having an interview conducted, they will both be able to immediately cease contact as well as have the opportunity to join the research activities again in the future. If a participant withdraws from the research after having an interview conducted, they will be able to follow the same procedure as a participant that withdraws before having an interview conducted. Additionally, they will have the opportunity to request that their previously collected interview data not be used in the research performed.

Your participation in this research study is entirely voluntary. You may want to discuss this study with your family and friends and your personal physician before agreeing to participate. If there are any words you do not understand, feel free to ask us. We will be available to answer

your current and future questions. Whether or not you provide your consent for participation in this research study will have no effect on your current or future relationship with the University of Pittsburgh.

Do you have any questions about this study?

We will always be available to address future questions, concerns or complaints as they arise. I certify that no research component of this protocol was begun until after this consent was obtained.

Please listen the following paragraph, where “I” represents you as the participant, and indicate whether or not you consent to participate in this study.

The above information has been explained to me and all of my current questions have been answered. I understand that I am encouraged to ask questions, voice concerns or complaints about any aspect of this research study during the course of the study, and that such future questions, concerns, or complaints will be answered by a qualified individual or by the investigators listed on the first page of this consent document at the email addresses given. I understand that I may always request that my questions, concerns, or complaints be addressed by a listed investigator. I understand that I may contact the Human Subjects Protection Advocate of the IRB Office, University of Pittsburgh (1-866-212-2668) to discuss problems, concerns, and questions; obtain information; offer input; or discuss situations that occurred during my participation. I voluntarily agree to participate in this research study.

Appendix E Institutional Review Board Approval



University of Pittsburgh *Institutional Review Board*

3500 Fifth Avenue
Suite 106
Pittsburgh, PA 15213
(412) 383-1480
(412) 383-1508 (fax)

MEMORANDUM

TO: Andrea L. Durst

FROM: Human Research Protection Office (HRPO)

DATE: July 12, 2018

SUBJECT: IRB# 1807003: Establish Infrastructure to Connect Underserved Populations to Genetic Services through a Toll-free Phone Line in New York-Mid Atlantic Consortium (NYMAC) Region

The above-referenced project has been reviewed by the Institutional Review Board. Based on the information provided, this project has been characterized as being an activity that does not meet the formal definition of research, according to the federal regulations at 45 CFR 46.102(d).

That is, the proposed activity is not a systematic investigation, including research development, testing, and evaluation, designed to develop or contribute to generalizable knowledge. Should the scope of this project change such that the definition of research is subsequently met, the investigator must notify the IRB immediately.

Given this determination, you may now begin your project.

Appendix F Histograms for Independent Variables in Analysis

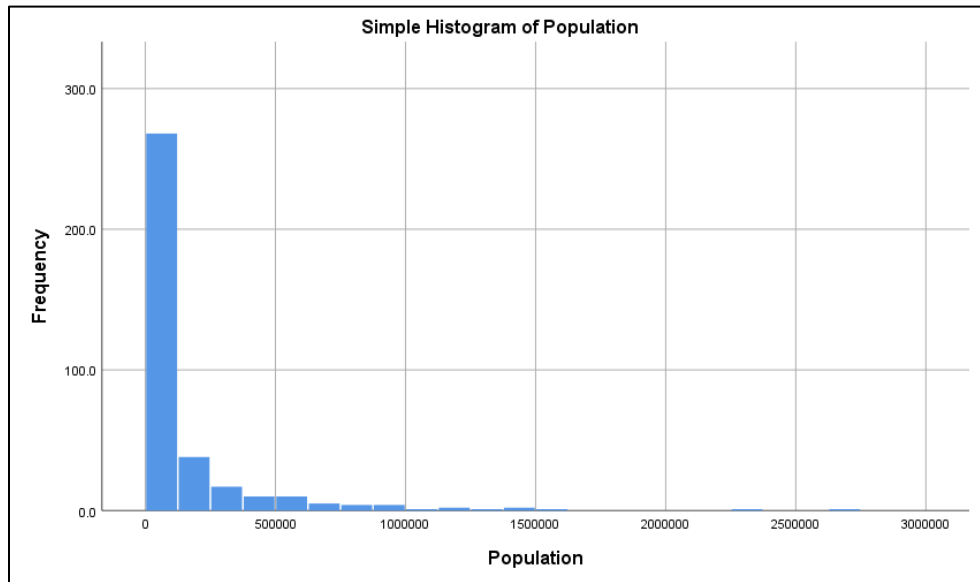


Figure 10 Histogram of Population by County.

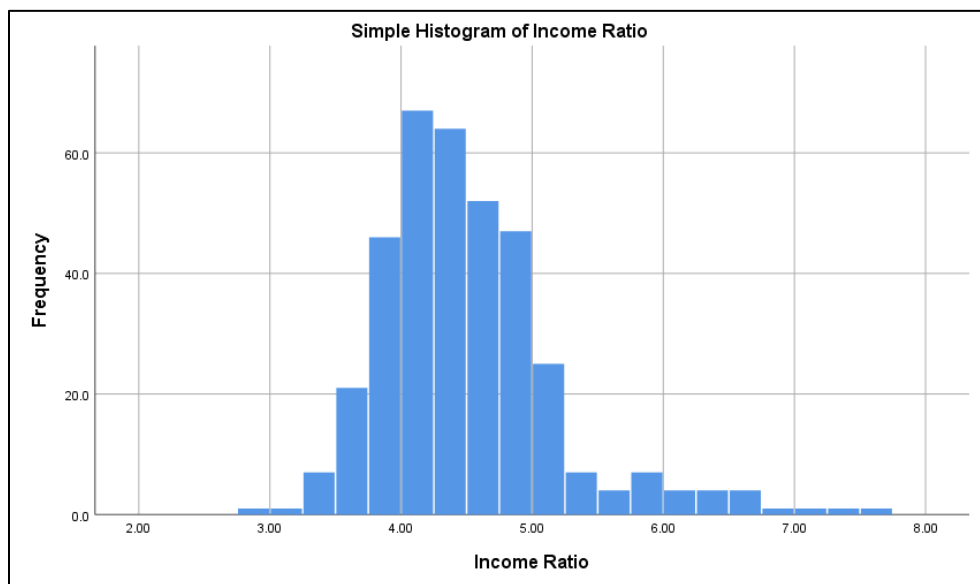


Figure 11 Histogram of Income Ratio by County.

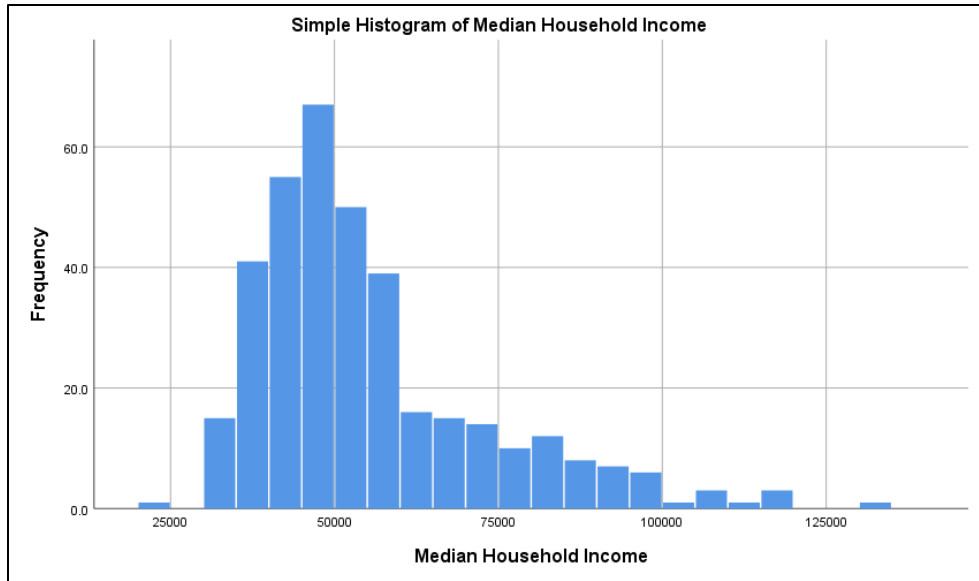


Figure 12 Histogram of Median Household Income by County.

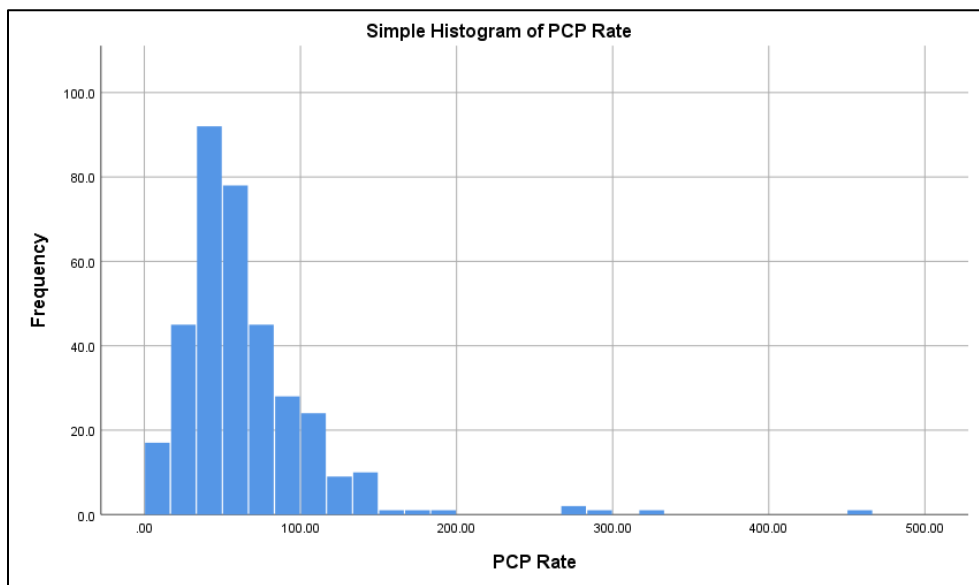


Figure 13 Histogram of PCP Rate per County.

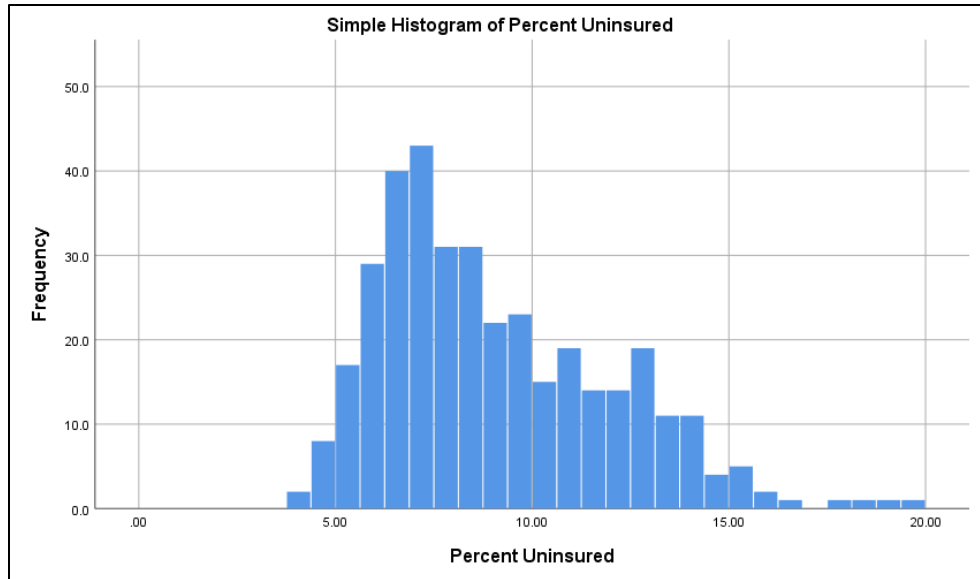


Figure 14 Histogram of Percent Uninsured by County.

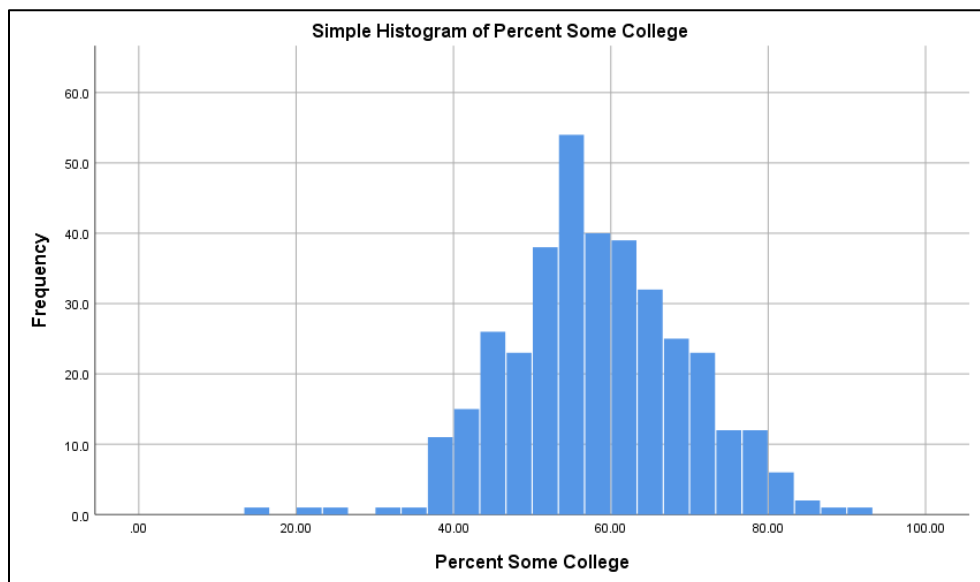


Figure 15 Histogram of Percent with Some College by County.

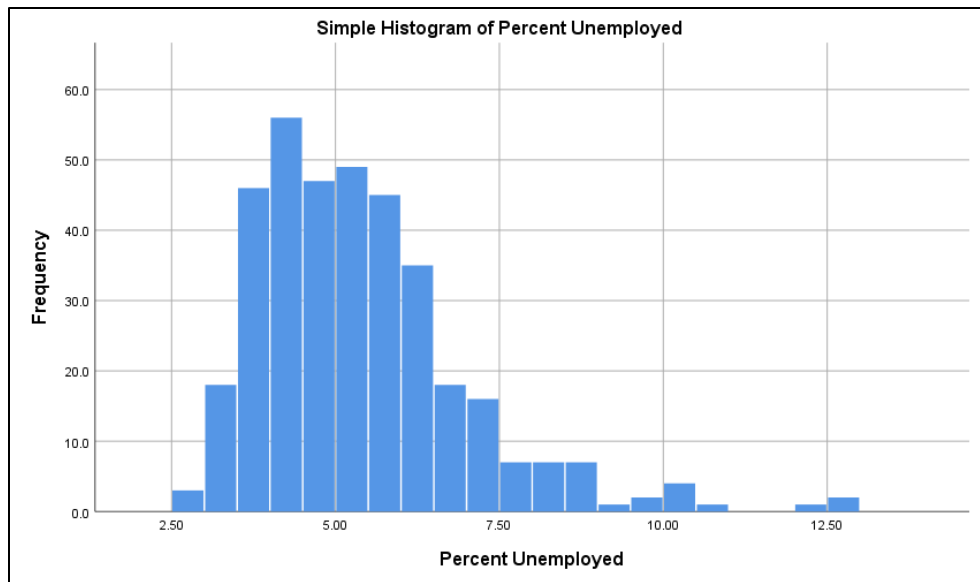


Figure 16 Histogram of Percent Unemployed by County.

Appendix G Scatter Plots Showing Correlation between Demographics, Clinics in Counties

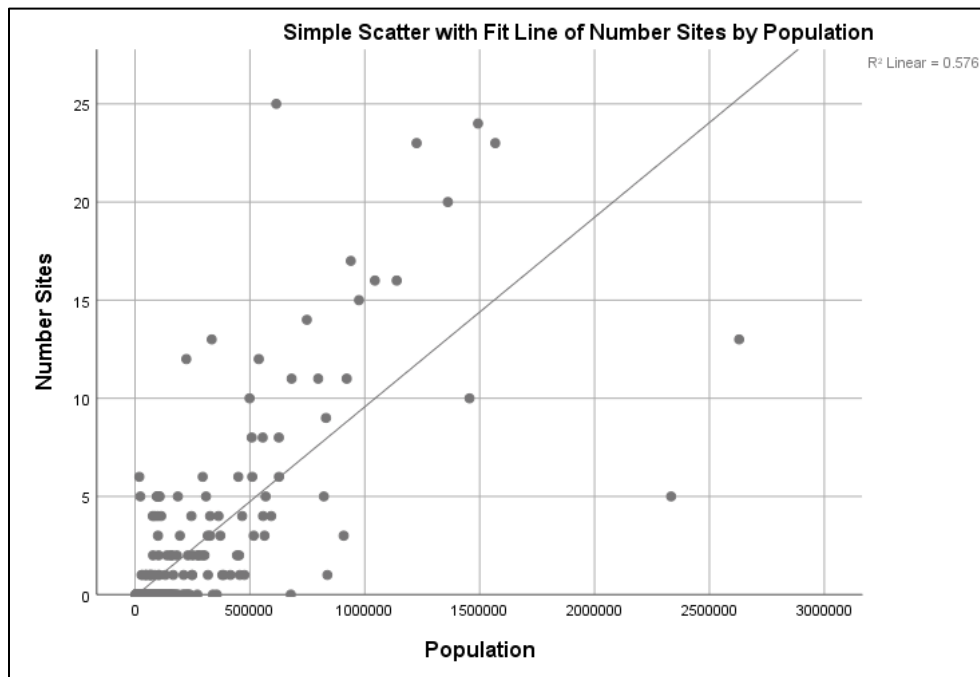


Figure 17 Scatter Plot Showing Correlation between Population and Number of Clinic Sites per County.

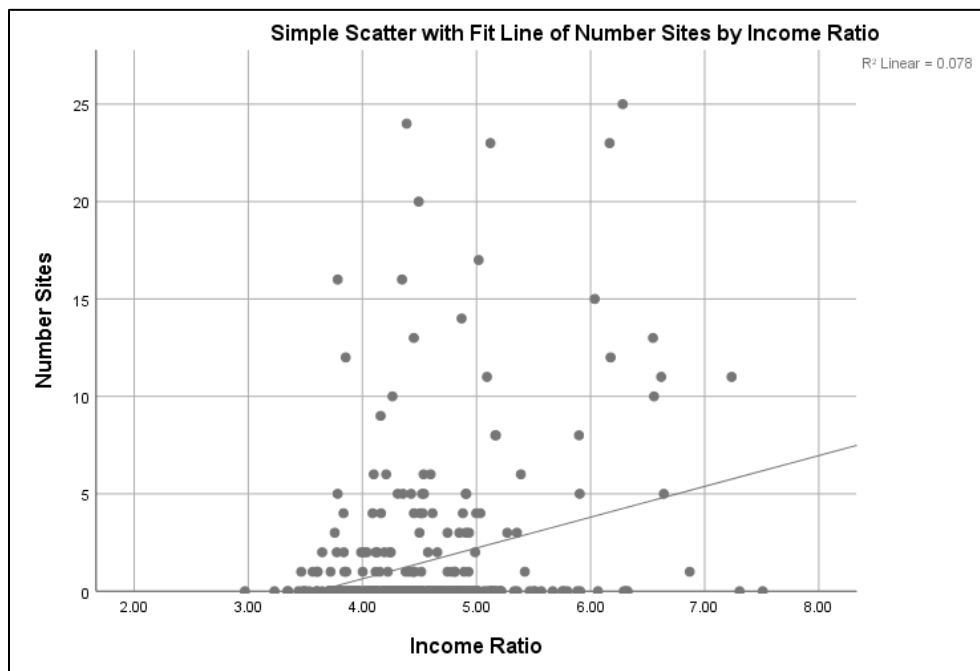


Figure 18 Scatter Plot Showing Correlation between Income Ratio and Number of Clinic Sites per County.

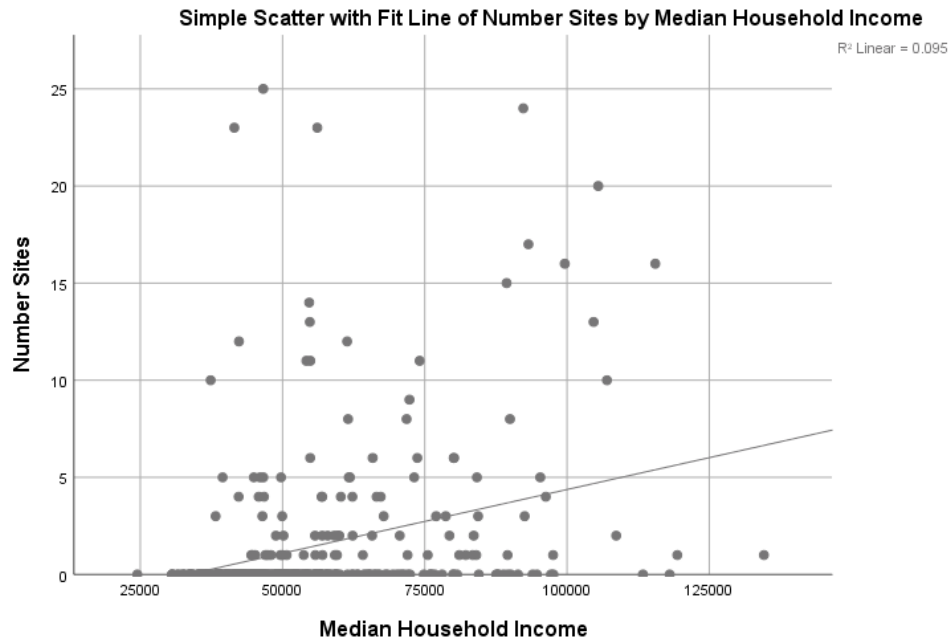


Figure 19 Scatter Plot Showing Correlation between Median Household Income and Number of Clinic Sites per County.

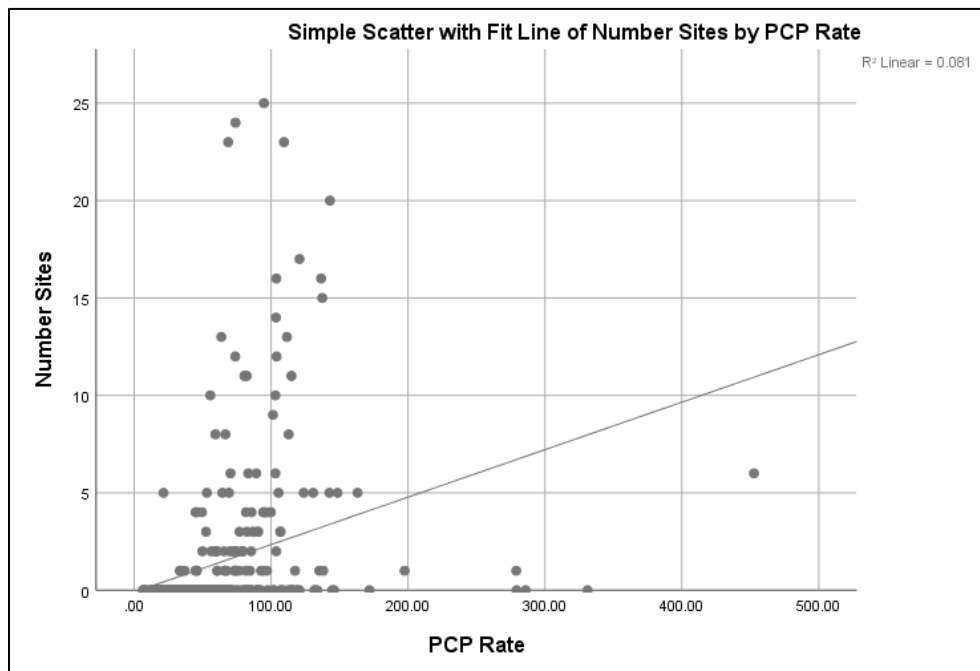


Figure 20 Scatter Plot Showing Correlation between PCP Rate and Number of Clinic Sites per County.

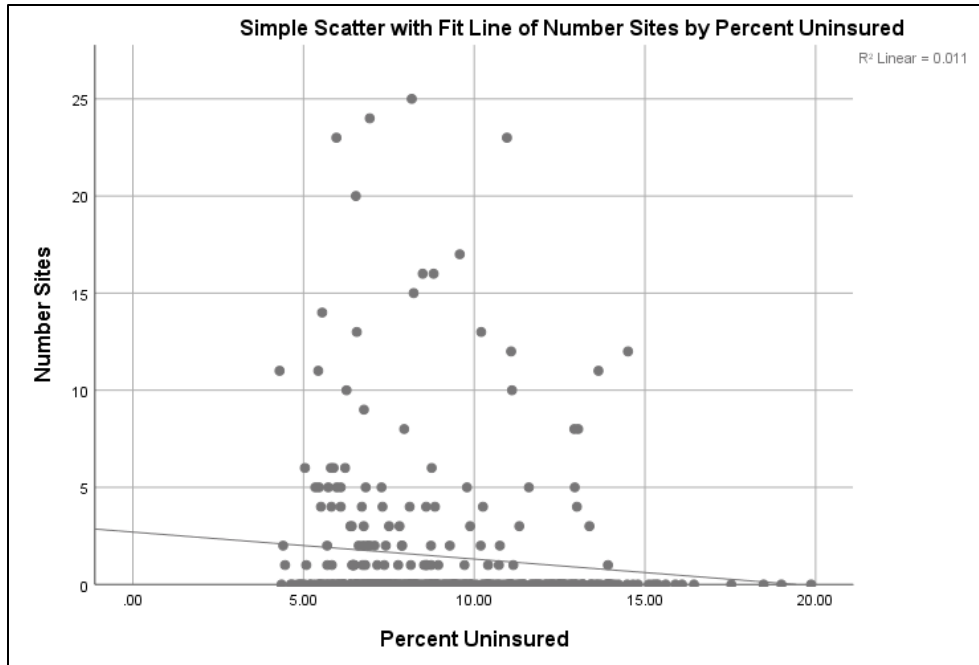


Figure 21 Scatter Plot Showing Correlation between Percent Uninsured and Number of Clinic Sites per County.

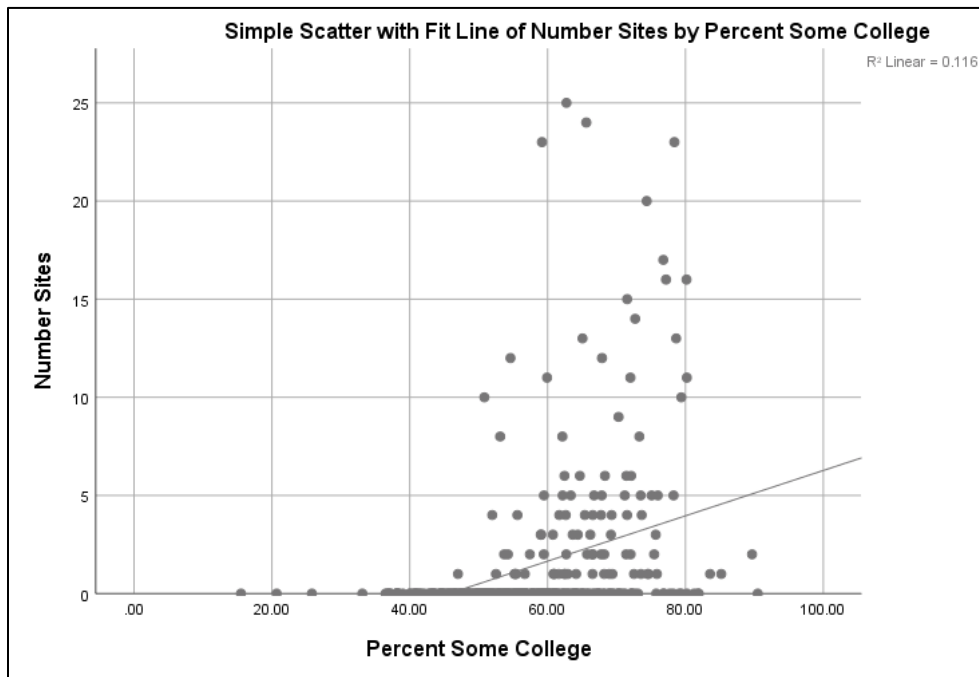


Figure 22 Scatter Plot Showing Correlation between Percent with Some College and Number of Clinic Sites per County.

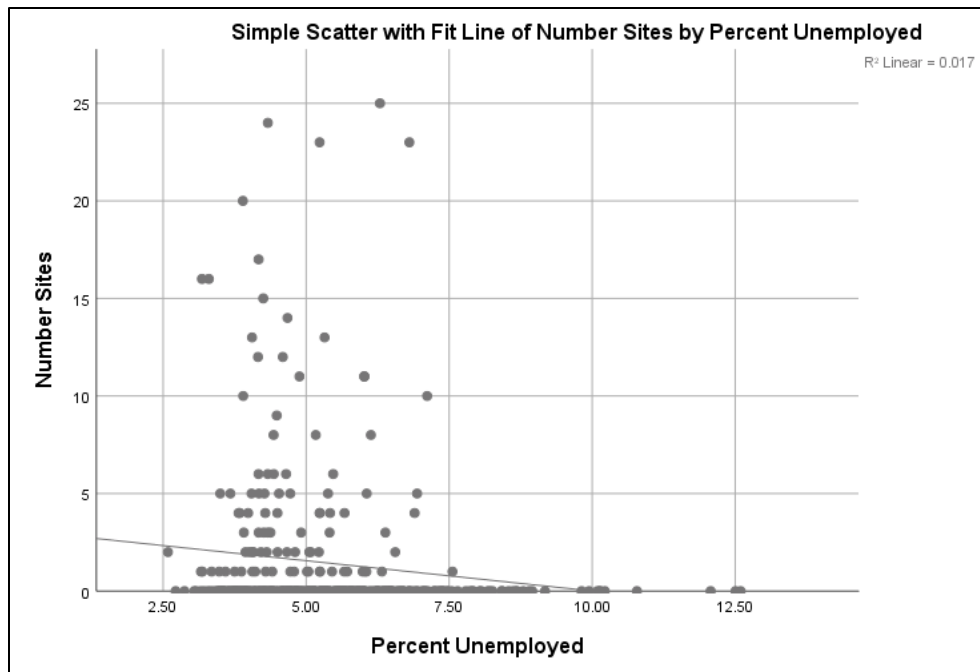


Figure 23 Scatter Plot Showing Correlation between Percent Unemployed and Number of Clinic Sites per County.

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